EPOS 2017
43rd Annual Meeting of the European Paediatric Ophthalmological Society
31 August - 2 September 2017
Institute of Mathematics, Oxford, UK

Programme

Local Hosts
Göran Darius Hildebrand (Scientific Organiser)
Raymond JF Lobo
Manoj Parulekar

https://www.epos-focus.org/meetings
http://www.epos2017.org
European Paediatric Ophthalmological Society (EPOS)
Topic and location of previous annual meetings

2001 - 2017

2017 Oxford, UK
Hereditary retinal dystrophies: from genetics to gene therapy

2016 Zürich, Switzerland
Paediatric Neuro-ophthalmology

2015 Saint Petersburg, Russia
Innovations in Paediatric Ophthalmology

2014 Barcelona, Spain
Paediatric Cataracts & Paediatric Glaucoma

2013 Leiden, The Netherlands
Paediatric Ophthalmic Tumours

2012 Uppsala, Sweden
Developments in Paediatric Ophthalmology

2011 Thessaloniki, Greece
Visual Impairment in Childhood

2010 Bad Nauheim, Germany
New Challenges in Paediatric Ophthalmology

2009 Paris, France
Perinatal Ophthalmology

2008 Leuven, Belgium
The Eye in Systemic Disease

2007 Portorož, Slovenia
Paediatric Electrophysiology and Psychophysics

2006 Vilamoura, Portugal
Pediatric Neuro-Ophthalmology

2005 Warszaw, Poland
Advances in the Surgical Treatment of Pediatric Eye Diseases

2004 Manchester, UK
Developmental Genes and the Eye

2003 Regensburg, Germany
Gene Therapy and other Modern Therapeutic Approaches in Paediatric Retinal Degenerations

2002 Figueras Daveo, Portugal
Dysmorphology of the Eye and Orbit

2001 Regensburg, Germany
Trends in Paediatric Ophthalmology

1996 Valencia, Spain
Neuro-ophthalmology

1995 Cambridge, UK
Dysmorphology and the Eye

1994 Regensburg, Germany
Teratology and the Eye

1993 Cambridge, UK
Phacomatoses

1992 Oxford, UK
Retinal Dystrophies

1991 Sandbjerg, Denmark
Multiply handicapped and the Ophthalmologist

1990 Oxford, UK
Anomalies of the Anterior Segment

1989 Bruges, Belgium
Retinopathy of Prematurity

1988 Oxford, UK
Genetic Diseases of the Cornea

1987 Geneva, Switzerland
Neuro-Ophthalmology

1985 Oxford, UK
Hearing and the Eye

1983 Amsterdam, The Netherlands
Genetics and Ophthalmology

1982 Oxford, UK
Ocular and Adnexal Tumours in Childhood

1981 Ghent, Belgium
Genetics and Ophthalmology

1980 Geneva, Switzerland
Retinal Disease in Childhood

1979 Oxford, UK
Visual Development in Childhood Normal and Abnormal

1978 Freiburg, Germany
Genetics and Ophthalmology

1977 Oxford, UK
Nystagmus

1976 Nijmegen, The Netherlands
Cataract in Childhood

1975 Copenhagen, Denmark
Visually Handicapped Children
The Ophthalmologist’s Responsibility

1974 Oxford, UK
Visual Function in Childhood

1973 Oxford, UK
Retinitis Pigmentosa

European Paediatric Ophthalmology Group (EPOG) 1973 - 2000

2000 Cambridge, UK
Retinal Dystrophies

1999 Strasbourg, France
Multisystem Disease and the Eye

1998 Dublin, Ireland
Metabolic Diseases of the Eye

1997 Cambridge, UK
Neonatal Ophthalmology

1996 Valencia, Spain
Neuro-ophthalmology

1995 Cambridge, UK
Dysmorphology and the Eye

1994 Regensburg, Germany
Teratology and the Eye

1993 Cambridge, UK
Phacomatoses

1992 Oxford, UK
Retinal Dystrophies

1991 Sandbjerg, Denmark
Multiply handicapped and the Ophthalmologist

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1977 Oxford, UK
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1976 Nijmegen, The Netherlands
Cataract in Childhood

1975 Copenhagen, Denmark
Visually Handicapped Children
The Ophthalmologist’s Responsibility

1974 Oxford, UK
Visual Function in Childhood

1973 Oxford, UK
Retinitis Pigmentosa
COMMITTEE

Host Committee
Göran Darius Hildebrand (Chairman and Scientific Programme Organiser) UK
Raymond JF Lobo UK
Manoj Parulekar UK

President
Nikolas Ziakas GREECE

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Veit Strum SWITZERLAND

Secretary
Eva Larsson SWEDEN

Board Members
Catherine Cassiman BELGIUM
Anne Cees Houtman BELGIUM
Sandra Valeina LATVIA
Göran Darius Hildebrand UK
Dear Colleagues and Friends,

It is my pleasure to welcome you to the 43rd Annual Meeting of the European Paediatric Ophthalmology Society (EPOS) in Oxford, UK. This year the meeting is hosted by Dr. Göran Darius Hildebrand and his team Manoj Parulekar and Raymond Lobo. They prepared an excellent program on the central theme “Hereditary Retinal Dystrophies: from Genetics to Gene Therapy”. It is a very exciting topic and, I believe, many new research data and clinical developments will be presented. There will also be interesting sessions on ROP, Oncology, Orbit and Craniofacial disorders, Cataract and Neuroophthalmology.

Several outstanding experts in the field have accepted the invitation to update us on their special interest. The program includes 27 invited lectures by international speakers from Europe and the USA. Apart from the lectures many participants will present their work covering a broad spectrum of topics. This year there will be 18 free papers, 40 rapid fire presentations and 79 posters. As always, best paper, rapid fire and poster presentations, for presenters under 35 years of age, will be awarded at the end of the meeting. I would like to point out that this year for the first time all accepted abstracts will be published online in the September - October issue of the European Journal of Ophthalmology (EJO).

As the main frame of EPOS is the exchange of new scientific ideas and experience in the field of paediatric ophthalmology, sufficient time for profound discussion in a friendly atmosphere is scheduled. This year the meeting will be attended by more than 220 delegates representing countries from virtually all over the world. We especially welcome our friends and colleagues from outside Europe.

Several exhibitors generously supported us again this year. I would like to express our sincere gratitude for their significant efforts and encourage all participants to visit the technical exhibit during coffee and lunch breaks. Finally, I would like to thank Agenda, the conference organizer, for their valuable help.

Our local hosts and their team prepared a meeting where science can be combined with culture and pleasure. We thank them for all their efforts. I wish you a fruitful meeting and hope that new collaborations, friendships and ideas will originate from our stay in historic Oxford.

Yours Sincerely,

Nikolas Ziakas
President of EPOS
Dear Friends and Colleagues,

I am delighted to welcome you to the historic city of Oxford for the 43rd Annual Meeting of the European Paediatric Ophthalmological Society!

The inaugural annual meeting of EPOS (it was not yet called this) took place in Oxford in 1973 and was dedicated to the condition Retinitis Pigmentosa. Like then, this year’s main focus will be on Hereditary Retinal Dystrophies: From Genetics to Gene Therapies, but also cover ROP and other retinal disorders, ocular oncology, craniofacial and orbit, cataract and neuro-ophthalmology.

I am very happy that we will welcome to our meeting paediatric ophthalmologists, neuro- and craniofacial surgeons, residents, orthoptists, optometrists and researchers in the field of ophthalmology, genetics and the neurosciences from across Europe and the rest of the world.

Oxford - and its younger cousin Cambridge - are the oldest Universities in the English-speaking world. Though the exact foundation date is not known, evidence suggests that university teaching started as early as 1096 and medicine has continuously been taught at Oxford since somewhere between 1220 and 1255. The University has educated many notable alumni, including 28 Nobel laureates, 27 Prime Ministers of the United Kingdom, over 50 Olympic medal winners and, it is said, 12 Saints!

The Conference will take place in the University’s spacious conference facilities at the brand-new Institute of Mathematics, located only a few minutes of walk away from the historic town centre and its venerable Colleges. The Institute’s immediate neighbour to the left is the former Radcliffe Infirmary, where penicillin underwent its first clinical testing on patients in 1941 following its successful isolation and purification in Oxford by Lord Florey and his team.

A highlight of our meeting will be gifting a Lifetime Achievement Award to the Society’s former President Prof Birgit Lorenz from Giessen, Germany.

I believe a most valuable aspect of a friendly meeting such as EPOS is the opportunity to meet colleagues and engage with them directly in- and outside of the Conference in the beautiful historic surroundings of the city. This should add tremendously to the academic experience.

In addition to the scientific programme, we have prepared what we hope will become a memorable social event as well. This year’s meeting will start with a Welcome and Social Reception between 18.30-20.30 on Thursday 31 August in the Master’s Garden of Christ Church College. You are welcome to bring your family and friends!

Due to the limited seating capacity of the Colleges and the large number of delegates this year, based on a first come, first serve principle, we have subdivided the delegates into two groups to be able to offer one dinner to everyone either Friday or Saturday evening in one of two of Oxford’s most beautiful Dining Halls: Balliol and Christ Church. Balliol College has existed as a community of scholars on its present site without interruption since about 1263. This token, it is said to be the oldest college in Oxford. Balliol will be the dinner venue for group 1 on Friday evening. Christ Church College stands out for its size, the beauty of its buildings, and its welcoming atmosphere. Saturday night we will be in the historic Dining Hall of Christ Church College, serving briefly as Parliament in the 17th century and familiar to many from the Harry Potter films!

I would like to warmly thank all delegates and speakers for coming from many parts of the world and our sponsors for their valuable financial support. No such conferences can be organised without thousands of emails and requests and a tireless team: I would like to thank the President, Prof Nikolas Ziakas, and the rest of the EPOS Board for all their valuable advice and support over the last 2 years to me and my local co-hosts Raymond Lobo and Manoj Parulekar. I finally need to thank our conference organiser Agenda for their enormous help.

I wish you a most rewarding three days to study and to meet old and new friends in Oxford!

Yours Sincerely,

Göran Darius Hildebrand

Scientific Programme Organiser and Chairman,
Local Host Committee
‘The Building of the Athenian Acropolis’ was an expression of the Promethean Spirit of discovery and advancement that befits a scientific conference

(detail from painting by Ernst Wilhelm Hildebrand, Academy of Fine Arts, Berlin, 1882)
GENERAL INFORMATION

Venue
Mathematical Institute
University of Oxford
Andrew Wiles Building
Radcliffe Observatory Quarter
Woodstock Road
Oxford
OX2 6GG

Insurance
Delegates are advised to take out travel insurance to cover medical expenses, accidents, loss, etc. No responsibility will be accepted by the Congress Organisers.

Presentation Upload, Registration & Opening Hours
Thursday 31st August 16.00 – 18.00
Friday 1 September 07.00 – 18.00
Saturday 2 September 07.00 – 18.00

CME Credits
The 43rd Annual Meeting of the European Paediatric Ophthalmological Society is accredited by the European Accreditation Council for Continuing Medical Education (EACCME) to provide the following CME activity for medical specialists. The EACCME is an institution of the European Union of Medical Specialists (UEMS), www.uems.net.

The 43rd annual meeting of the European Paediatric Ophthalmological Society is designated for a maximum of (or “for up to”) 12 hours of European external CME credits. Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

Through an agreement between the European Union of Medical Specialists and the American Medical Association, physicians may convert EACCME credits to an equivalent number of AMA PRA Category 1 Credits™. Information on the process to convert EACCME credit to AMA credit can be found at www.ama-assn.org/go/internationalcme.

Live educational activities, occurring outside of Canada, recognized by the UEMS-EACCME for ECMEC credits are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of The Royal College of Physicians and Surgeons of Canada.

WIFI
How to connect to the Cloud WiFi
1. Switch on your smartphone, tablet or laptop and check that WiFi is enabled.
2. Select ‘_The Cloud’ from the available network list.
3. Open your internet browser - the venue landing page will appear. If it does not, type in bbc.co.uk
4. If it is your first time using The Cloud WiFi network, follow the simple one-time registration process by sharing some simple details.
5. Once registered you can access the internet via The Cloud

Uploading of Presentations
Participants are advised to upload their presentation well in advance of their session to the computer in the speaker room. It will not be possible to use a personal laptop. Speakers can upload their presentations from Thursday afternoon at the Institute of Mathematics.

Presentation time (unless indicated otherwise) will be 17 minutes for invited lectures, 5 minutes for free papers and 2 minutes for rapid fire (RF) presentations. Time for discussion will be 3 minutes for invited papers and free papers and 1 minute for RF presentations. You are kindly requested not to exceed the assigned time.

Posters
Posters will be displayed for 1 day. The day and order of display has been allocated and can be found at the beginning of the poster section. You will find the pre-numbered poster boards in the Institute of Mathematics.

Grants
At the end of the annual meeting grants for best presentation (free paper and rapid fire) and poster will be awarded. Any registered member participating in the annual meeting aged 35 years or less may apply for this award which will be decided upon by the Board of the Society. The actual presentation as well as the response to questions about the paper or poster will be taken into account.

Parking
There is no parking at the Institute of Mathematics. Delegates are advised to use public transport or to walk.

Map of Oxford
Please see page 97
SCIENTIFIC CONTENT
THURSDAY 31 AUGUST

16.00 – 18.00
Registration Open
Speaker Ready Room Open
Institute of Mathematics,
University of Oxford

18.45 – 20.30
Welcome and Social Reception
Master’s Garden,
Christ Church College, Oxford

FRIDAY 1 SEPTEMBER

08.45 – 10.30
Inherited Retinal Dystrophies
Chairpersons: John Marshall UK
Ingele Casteels BELGIUM

08.45 John Marshall UK (Keynote Speaker)
The Structure of the Retina and its Implications for Retinal Dystrophies

09.05 Marcus Fruttiger UK
Retinal development

09.25 Kristina Irsch USA
Adaptic optics

09.45 Free Paper and Rapid Fire Session
Chairpersons: Susie Downes UK
Kristina Irsch USA

09.45 Hans Ulrik Moller DENMARK
Which is the more difficult to diagnose - retinal dystrophies or corneal dystrophies?

09.53 Maria Toms, Dhani Tracey-White, Rose Richardson, Thomas Burgoyne, Mariya Moosajee UK
KCNJ13-related severe early onset retinal degeneration associated with a mitochondrial aging pathophysiology

10.01 Charlotte Krujt, Gerard de Wit, Bergen Arthur, Nicoline Schalij-Delfos, Maria van Genderen THE NETHERLANDS
Phenotypic spectrum of albinism

10.09 Efthymia Prousali, Asimina Mataftsi, Nikolaos Ziakas, Andreas Fontalis, Anna-Bettina Haidich GREECE
Interventions to control myopia progression in children: An overview of systematic reviews and meta-analyses

10.14 Sibel Oto, Almila Sarıgül Sezenöz, İmren Akkoyn, Sezin Bayar Akça, Gürsel Yılmaz TURKEY
OCT findings and visual prognosis in children with unilateral myelinated retinal nerve fibers and high myopia

10.20 Sabine Defoort-Dhellemmes, Isabelle Drumare, Vasili Smirnov, Bernard Puech FRANCE
SD-OCT and FAF at onset of inherited maculopathy in children

10.23 Nathalie Voide, Francis L. Munier SWITZERLAND
Torpedo maculopathy: a multimodal analysis of a pediatric case series

10.26 Victoria Pueyo, Jose Ignacio Echeverria, Kim Plunkett SPAIN
Validity and usability of new stimuli for colour vision assessment in infancy

10.26 Annegret Dahlmann-Noor, Rachel Thomas, Michael Crossland, Hilary Unwin, Vijaya Gothwal UK
Assistive technology for children with low vision: the CREATE pilot trial (Children Reading with Electronic Assistance To Educate)
FRIDAY 1 SEPTEMBER

10.30 – 11.00
Coffee Break and Poster Exhibition

11.00 – 13.15
Inherited Retinal Dystrophies Continued

Chairpersons: Peter Charbel Issa UK
Anne Cees Houtman BELGIUM

11.00  Peter Charbel Issa UK
Clinical manifestations of inherited retinal dystrophies

11.20  Dorothy Thompson UK
Electrodiagnostic testing for inherited retinal dystrophies

11.40  Susie Downes UK
Genetic basis of retinal dystrophies

12.00  Robert MacLaren UK
Gene therapy for inherited retinal dystrophies

12.20  Free Paper and Rapid Fire Session
Chairpersons: Robert MacLaren UK
Catherine Cassiman BELGIUM

12.20  Irina Balikova, Casper De Jong, Sophie Walraedt, Hilde Deconinck, Elfride De Baere, Bart P. Leroy BELGIUM
Autosomal Recessive Bestrophinopathy in the pediatric population - clinical features and diagnostic challenges

12.28  Cameron Parsa, Aurélie Taylor USA
How to explain “flat” electroretinograms when patients with Leber's congenital amaurosis aren’t blind

12.36  Panagiotis Sergouniotis, Rachel Taylor, Neil Parry, Susmito Biswas, Jane Ashworth, Graeme Black UK
The diagnostic yield of clinical genetic testing in paediatric inherited retinal disease

12.44  Markus Preising, Ute Schneider, Hanno Jörm Bolz, Christoph Friedburg, Andrassi-Darida Monika, Melanie Jäger, Birgit Lorenz GERMANY
Genotype-phenotype correlation in inherited retinal diseases caused by biallelic CEP290 variations

12.52  Chadha Rasmee UK
10 year audit of The Paediatric Low Vision Aid Clinic at Oxford Hospital

12.55  Mohamed Adam Ali, Dhani Tracey-White, Matthew Smart, Andrew Webster, Mariya Moosajee UK
Combined nonsense-mediated decay inhibition and translational readthrough as a treatment approach for hereditary retinal dystrophies

12.58  Amandine Monnier, Sarah Michel, Olivia Zambrowski, Dominique Bremond Gignac, Matthieu Robert UK
Post infectious pigmentary retinopathies in infants

13.01  Nataly Osipova, Lyudmila Katargina, Ekaterina Denisova, Lyudmila Kogoleva, Oiga Novikova RUSSIA
Choroidal neovascularization secondary to Best vitelliform macular dystrophy: Is it rare?

13.04  Martina Jarc-Vidmar, Branka Stim-Kranjc, Polona Jaki-Mekjič SLOVENIA
Intravitreal anti-VEGF treatment for choroidal neovascularisation secondary to Best vitelliform macular dystrophy in a five year old child and his grandmother

13.07  Anna Majander, Eeva-Marja Sankila, Kristiina Vasara, Anna-Kaisa Haavisto, Sanna Seitsonen, Kristiina Avela FINLAND
A TULP1 mutation in Finnish patients with congenital nystagmus and early onset retinal dystrophy (EORD)

13.10  Vasily Smirnov, Isabelle Drumare, Sabine Defoort-Dhellemmes FRANCE
Hereditary retinal diseases diagnosed in infants with congenital vertical nystagmus

13.15 – 14.00
Lunch Break and Poster Presentation
FRIDAY 1 SEPTEMBER

14.00 – 15.45

ROP
Chairpersons:  
Eva Larsson SWEDEN  
Annegret Dahlmann-Noor UK

14.00  
Sir Peter Ratcliffe UK (Keynote Speaker)  
How cells sense and signal hypoxia

14.20  
Birgit Lorenz GERMANY (Keynote Speaker)  
Clinical presentation and classification of ROP

14.40  
CK Patel UK  
Clinical and surgical management of ROP

15.00 – 16.15  
Coffee Break and Poster Exhibition

16.15 – 17.00  
Other Retinal Disorders
Chairpersons:  
CK Patel UK  
Sandra Valeina LATVIA

16.15  
Martin Snead UK  
Paediatric Retinal Detachment

16.35  
Creig Hoyt USA (Keynote Speaker)  
Retinal haemorrhages in children

17.15 – 18.15  
EPOS General Assembly

19.45  
Conference Dinner at Balliol College Group 1

15.00  
Free Paper and Rapid Fire Session
Chairpersons:  
Birgit Lorenz GERMANY  
Gerd Holmström SWEDEN

15.00  
Annegret Dahlmann-Noor, Gillian Adams, Lucilla Butler, Wen Xing, Catey Bunce UK  
Findings from the national UK Retinopathy of Prematurity Treatment Study

15.08  
Lotta Gränse, Ulrika Kjellström, Sten Andrèasson SWEDEN  
New possibilities to examine small children and infants with unknown vision loss

15.16  
Gerd Holmstrom, Jonina Hreinsdottir, Ylva Kaul-Fredriksson, Claes von Hofsten, Kerstin Roander, Lena Westas-Hellstrom SWEDEN  
Prediction of ophthalmological problems in 6.5 year-old prematurely born children - preliminary results of a population-based, prospective study

15.24  
Erika Maka, Andrea Szigeti, Maria Bausz, Miklos Resch, Zoltan Zsolt Nagy HUNGARY  
Is ROP the worst abnormality what we can find in a preterm baby?

15.27  
Alena Gerasimenko, Victoria Krasilnikova BELARUS  
The macula zone status in cicatrical period of retinopathy of prematurity according to optical coherence tomography

15.30  
Alena Gerasimenko, Victoria Krasilnikova BELARUS  
Late retinal detachment in adult retinopathy of prematurity

15.33  
Malgorzata Kowalczyk, Robert Rejdak POLAND  
Autosomal dominant neovascular inflammatory vitreoretinopathy

15.36  
Gerard Reid, Marie O’Neill, Eibhlin McLoone UK  
Retinal injuries resulting from handheld laser devices in the paediatric population, A case series of 27 eyes

15.39  
Christina Gerth-Kahler, Lisa Weibel, Martin Theiler, Francis Munier SWITZERLAND  
Severe ocular complications in patients with macrocephaly-capillary malformation syndrome

15.42  
Teresa Pérez Roche, Esther Prieto, Olimpia Castillo, Javier Gutierrez, Marta Ortin, Diego Gutierrez SPAIN  
Age-related normative values for visual fixation measured by a digital device
07.00 – 18.00
Registration Open

08.00 – 09.00
Oncology Session
Chairpersons: Manoj Parulekar UK
Mandeep Sagoo UK

08.00 Manoj Parulekar UK
Retinoblastoma and its management

08.20 Mandeep Sagoo UK
Other intraocular tumours in childhood

08.40 Free Paper and Rapid Fire Session
08.40 Joana Providência, Guilherme Castela, Sónia Silva, Egidio Machado PORTUGAL
First 22-months results of retinoblastoma management in a national reference center

08.43 Parth Shah, Fariha Shafi, Maureen McCalla, Zoe Squires, Manoj Parulekar UK
Outcomes following enucleation for retinoblastoma

08.46 Lucia Derriman, Mohammad Ayoubi, Arinder Channa, Irina Gout, Abbas Toufeeq UK
Rare case of permanent, bilateral severe visual loss secondary to craniosynostosis in Alagille Syndrome

09.00 – 10.00
Craniofacial and Orbital Session
Chairpersons: Geoff Rose UK
Jonathan Norris UK

09.00 Greg Thomas UK
Ophthalmology in Craniofacial Disorders

09.20 Geoff Rose UK (Keynote Speaker)
Tales from the Children’s Toolbox

09.40 Göran Darius Hildebrand UK
Topical beta-blocker treatment for capillary hemangioma in infancy

10.00 – 10.30
Coffee Break and Poster Exhibition

10.30 – 11.10
Retinal Implant
10.30 Eberhart Zrenner GERMANY (Keynote Speaker)
The retinal implant: from idea to reality

11.10 – 12.40
Cataract and Lens Session
Chairpersons: Boris Malyugin RUSSIA
Göran Darius Hildebrand UK

11.10 Christopher Lloyd UK
Causes for childhood cataracts and ectopia lentis

11.20 Göran Darius Hildebrand UK
Cataract surgery in children

11.50 Boris Malyugin RUSSIA (Keynote Speaker)
Ectopia lentis and its surgical management

12.20 Marie-José Tassignon BELGIUM (Keynote Speaker)
The “Bag-in-the-lens” implant in pediatric cataract surgery

12.40 – 13.40
Lunch Break and Poster Exhibition

13.40 Free Paper and Rapid Fire Session
Chairpersons: Nikolas Ziakas GREECE
Marie-José Tassignon BELGIUM

13.40 Ameenat Lola Solebo, Phillippa Cumberland, Jungnoo Rahi, BCCIG British Isles Congenital Cataract Interest Group UK
Visual outcomes five years following congenital and infantile cataract surgery with and without primary IOls: findings from the IOLunder2 study

13.48 James Neffendorf, Elizabeth Hawkes, Vicki Sandford, Göran Darius Hildebrand UK
Safety and outcome of paediatric cataract surgery

13.56 Alf Nyström, Nawal Almarzouki, Gunilla Magnusson, Madeleine Zetterberg SWEDEN
Bag-in-the-lens intraocular lens - well suited for use in children

14.04 Alf Nyström, Birgitte Haargaard, Gunilla Magnusson, Annika Rosensvärd, Kristina Tornqvist SWEDEN
Glaucoma after cataract surgery in childhood - a report on 8 years of register based follow-up

14.12 Annegret Dahlmann-Noor, Daniel Moritz, Adam Dubis, Ana Quartiilo, Peng Khaw, Maria Theodorou UK
Lensectomy reduces Schlemm canal diameter - a mechanical hypothesis of post-lensectomy glaucoma
SATURDAY 2 SEPTEMBER

14.20  Dimitrios Giannoulis, Eirini Kostopoulou, Angeliki Chroniati, Asimina Mataftsi, Nikolas Ziakas 
GREECE  
Simultaneous bilateral cataract surgery with IOL implantation in pediatric patients

14.23  Parth Shah, Laura Ramm, Mary Awad, Clare Dewsbury, Manoj Parulekar 
UK  
Orthoptist-led aphakia clinics

14.26  Annegret Dahlmann-Noor, Moritz Daniel, Adam Dubis, Ana Quartilho, Peng Khaw, Maria Theodorou 
UK  
Dynamic changes of iridocorneal angle morphology during accommodation in healthy children

14.29  Annegret Dahlmann-Noor, Moritz Daniel, Adam Dubis, Ana Quartilho, Peng Khaw, Maria Theodorou 
UK  
Scheimpflug lens densitometry: an objective measure of cataract severity in children

14.32  Esther Papamichael, Sahar Parvizi, Kunal Gadhvi, Preadeepan Vetpillai, Melanie Hingorani 
UK  
Is Ciclosporine 0.1% useful in children with severe ocular surface inflammation?

14.40 – 15.10  
Coffee Break and Poster Exhibition

15.10 – 17.15  
Neuro-Ophthalmology Session

Chairpersons:  Cameron Parsa  USA  
John Elston  UK

15.10  David Taylor  UK (Keynote Speaker)  
Papilloedema or not?

15.30  Cameron Parsa  FRANCE  
Neuro-ophthalmic manifestations of brain tumours in children

15.50  Free Paper and Rapid Fire Session

Chairpersons:  David Taylor  UK  
Creig Hoyt  USA

15.50  Guy Mole, Rachel Edminson, Caroline Hopper, Caroline Hildebrand  UK  
Paediatric tumours and their ophthalmic management in the paediatric ophthalmology clinic at a tertiary paediatric oncology centre

15.58  Mervyn Thomas, Gail Maconachie, Viral Sheth, Rebecca McLean, Irene Gottlob  UK  
A novel diagnostic next generation sequencing panel for infantile nystagmus

16.06  Rushmia Karim, Rebecca Jones, Dilys Oladiwura, Annegret Dahlmann-Noor, Andrew Sawczenko  UK  
Diagnostic pathway of optic atrophy in the paediatric population

16.09  Marita Andersson Grönlund, Lovisa Mybeck, Susann Andersson  SWEDEN  
Imaging of the retina and optic nerve using optical coherence tomography in adolescents with surgically treated hydrocephalus

16.12  Matthieu Robert, Diem-Trang Nguyen, Gilles Martin, Amandine Pon-Monnier, Olivia Zambrowski  FRANCE  
The usefulness of visual electrophysiology in pendular nystagmus in infancy

16.15  Martin Wasik, Rachel Edminson, Goran Darius Hildebrand  UK  
The neuro-ophthalmic complications of childhood medulloblastoma and its treatment

16.18  Zuzana Sipkova, Guy Mole, Göran Darius Hildebrand  UK  
Neuro-ophthalmic features of paediatric diencephalic tumours

16.21  Kavita Aggarwal, Fabienne Fierz, Shaun Wilson, Kate Wheeler, Göran Darius Hildebrand  UK  
Ophthalmic complications of proton beam radiotherapy for paediatric patients with intracranial and orbital tumours

16.24  Nathalie Voide, Matthieu Robert  SWITZERLAND  
The Heimann-Bielschowsky phenomenon: a pediatric case series

16.27  Krishanthy Sornalingam, Ahmed Javed, Panagiotis Sergouniotis, Tariq Aslam, Jane Ashworth  UK  
Retinal imaging as an objective measure of ocular disease in Mucopolysaccharidosis

16.30  Susann Andersson, Johanna Norström, Marita Andersson Grönlund  SWEDEN  
Vision and visual perception in adolescents with hydrocephalus - a long term follow-up

16.33  John Elston  UK  
Optic nerve sheath decompression surgery

16.53  Shailendra Magdum  UK  
Neurosurgery for raised ICP

17.20 – 18.30  
Awards, Presentation for next year and Closing Remarks

19.45  Conference Dinner at Christ Church College  Group 2
LECTURE, FREE PAPER & RAPID FIRE ABSTRACTS
The Structure of the Retina and its Implications for Retinal Dystrophies

John Marshall

UCL Institute of Ophthalmology, London - UK

A brief account of retinal anatomy will be given with emphasis on the structural differentiation between fovea, fovea, macular and equatorial retina together with common misinterpretations. The implications of differential structures in these topographic locations will be discussed in relation to disease processes occurring in specific cellular systems. Given that the majority of the dystrophies result from genetic lesions in the outer retina the dynamic processes involved in the maintenance of the outer blood retinal barrier and the sustained survival of photoreceptor cells will be highlighted.
Retinal development

Marcus Fruttiger

UCL Institute of Ophthalmology, London - UK
Adaptive Optics (AO) refers to a technique to compensate for distortions caused by optical aberrations in the media between the object being imaged and the camera. It was originally developed for use in astronomical telescopes to compensate for optical distortions induced by the inhomogeneous earth atmosphere. It has since evolved to become a powerful clinical tool in ophthalmology. In the eye, a “wavefront sensor” (aberrometer) measures the distortion of transmitted light that is induced by inhomogeneities within the cornea and crystalline lens. The light is then “undistorted” after being reflected by a mirror that is suitably deformed. AO thus enables imaging of the ocular fundus with unprecedented resolution in vivo, such as revealing defects of nerve fiber layer, alterations in blood vessel walls, warps and deformations of the pores of the lamina cribrosa, and individual photoreceptors. AO by itself does not provide an image; rather an AO subsystem is incorporated into an existing imaging device. AO subsystems have thus far been successfully integrated into three ophthalmic imaging devices: fundus cameras, scanning laser ophthalmoscopes, and the OCT device. This lecture will introduce the basic optical principles of AO and illustrate its value with state-of-the-art clinical examples.
**Which is the more difficult to diagnose - Retinal dystrophies or corneal dystrophies?**

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*Aarhus University Hospital, Haarhaus - Denmark*

**Introduction:** as this years theme is retinal dystrophies I want to put into perspective which dystrophies puzzle the paediatric ophthalmologist most.

**Methods:** based on 25 years of interest and being part of the International committee of classification of corneal dystrophies I want to answer the question above.

**Results:** Most eye docs think the answer is corneal - the answer is retinal dystrophies.

**Conclusion:** corneal dystrophies may be quite straight forward if you approach them correctly.

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**KCNJ13-related severe early onset retinal degeneration associated with a mitochondrial aging pathophysiology**

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*Moorfields Eye Hospital NHS Foundation Trust, London - UK*

**Introduction:** Mutations in KCNJ13, which encodes a defective inwardly-rectifying K+ channel (Kir7.1) expressed in the apical membrane of RPE, cause Leber Congenital Amaurosis (LCA). Here, we characterise the kcnj13-/- zebrafish (p.F168L), to investigate the retinal pathophysiology.

**Methods:** Retinal structure and cone mosaic regularity were examined using histology, electron microscopy and SD-OCT from 3-12 months. Vision was assessed using the optokinetic reflex assay. Müller cell activation (GFAP immunostaining), cell death assays and western blot of heat shock protein 60 (hsp60) were undertaken. Mitochondrial ATP/energy production was measured using a luciferin-luciferase assay. qRT-PCR of mitochondrial genes; polg, nd1 (copy number), fis1 (fission), opa1 (fusion), pgk1 (glycolysis), ldha (anaerobic respiration), sod1/2 (reactive oxygen species, ROS).

**Results:** Electron microscopy uncovered an expansion of mitochondria and phagosomes in the RPE at 6 months, prior to any histological changes, confirmed by qRT-PCR showing increases in nd1, polg, fis1, opa1 and pgk1. Significant retinal thinning was noted at 12 months, along with disruption of the cone mosaic regularity and decline of visual function. Furthermore, Müller cell activation, reduced ATP production, increased levels of hsp60 and cell death were detected at both 6 and 12 months.

**Conclusion:** Dysfunction of the Kir7.1 channel results in significant mitochondrial changes within the RPE and photoreceptors. This is likely to be a compensatory response to metabolic demand and cellular stress. Similar processes have been seen in models of age-related macular degeneration. This study provides an insight into the disease mechanisms of LCA and other retinal degenerations, highlighting mitochondrial behaviour as a potential therapeutic target.
Phenotypic spectrum of albinism

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Introduction: Albinism is a genetically determined disorder caused by defects in the melanin-biosynthesis pathway. Ophthalmic characteristics include reduced visual acuity (VA), nystagmus, iris translucency, foveal hypoplasia, and optic nerve misrouting. In misrouting a majority of the optic nerve fibers cross at the chiasm. Based on molecular analysis, albinism can be divided in several subtypes. To demonstrate the variability of albinism we made an overview of the phenotypic spectrum of a large cohort of albinism patients.

Methods: We retrospectively investigated clinical, genetic, and electrophysiological data of 519 patients from the databases of Bartiméus (N=452), LUMC (N=40), and AMC (N=27), from 1982 until 2016. Clinical data included VA, nystagmus, iris translucency, fundus pigmentation, and foveal hypoplasia. We graded iris translucency from 1 (minimal punctuate translucency) to 4 (complete translucency) and fundus pigmentation from 1 (normally pigmented fundus) to 3 (transparent pigment epithelium). Based on SD-OCT images we graded foveal hypoplasia from 0 (no hypoplasia) to 4 (severe hypoplasia). Of the 167/519 patients that were genetically tested the diagnosis was confirmed in 76.6% (128/167).

We included patients if they met one of the following criteria:
- A confirmed molecular diagnosis of albinism;
- Apparent hypopigmentation of skin and hair and/or grade 2 or 3 fundus hypopigmentation, accompanied by at least two of the following characteristics: iris translucency, misrouting, foveal hypoplasia, nystagmus;
- Iris translucency and at least two of the following ophthalmic characteristics: misrouting, foveal hypoplasia, nystagmus.

Results: In this large cohort of patients with albinism, VA ranged from 1.3 logMAR to -0.1 logMAR (mean 0.56 logMAR). Nystagmus was absent in 41/514 (8%) and iris translucency was absent in 45/409 (11%). Fundus pigmentation was normal in 20/436 (4.6%). In 1/142 (0.7%) foveal hypoplasia was absent. Misrouting could not be demonstrated in 50/286 (17.5%).

Conclusion: The results of this study demonstrate the phenotypic heterogeneity of albinism. Clinical diagnosis may be difficult to establish, since 35% of our patients had a VA < 0.3 logMAR and no ophthalmic characteristic was consistently present in all patients. This implies that a complete ophthalmic examination, including VEP, as well as mutation analysis, is necessary to either confirm or reject the diagnosis, especially in patients with very mild disease.

Interventions to control myopia progression in children: An overview of systematic reviews and meta-analyses

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Introduction: Myopia is a common visual disorder with increasing prevalence among developed countries of the world. The purpose of this study was to conduct an overview of systematic reviews and meta-analyses in order to identify and appraise level 1 evidence regarding the management of myopia progression in children and adolescents.

Methods: Literature search was conducted in MEDLINE, EMBASE, The Cochrane Database of Systematic Reviews (CDSR) and Database of Abstracts of Reviews of Effects (DARE), to January 15th 2017. Individual study methodological quality and quality of evidence were assessed by 2 independent authors using the ROBIS tool and GRADE rating, respectively. Recently published primary studies were identified and discussed. A citation matrix was generated and the corrected covered area (CCA) was estimated, in order to identify overlapping primary studies. Meta-biases and measures of heterogeneity were reported and cases of dual co-authorship were identified.

Results: Out of 1975 non-duplicate records, 21 full texts were considered relevant and retrieved, of which 14 systematic reviews and meta-analyses and 3 new primary studies were included. These comprised a total of 91 primary studies which were qualitatively synthesized. The overall risk of bias was judged as ‘low’ in ten included reviews, ‘unclear’ in two, and ‘high’ in two studies. Quality of evidence was assessed as ‘high’ in five reviews, ‘moderate’ in seven, and ‘low’ in two reviews. Reported findings were concordant, suggesting that atropine eyedrops, orthokeratology and modern multifocal soft contact lenses demonstrate efficacy in myopia control. The corrected covered area (CCA) was calculated 7% and therefore considered slight with a low risk of skewed reporting. Very large (I²>75%) and large heterogeneity (I²>50%) were reported by two and four meta-analyses, respectively. Associated adverse events have prevented efficient interventions from becoming widely accepted for myopia treatment. The most common are photophobia, mydriasis, cycloplegia, rebound effect for atropine, and microbial keratitis for orthokeratology.

Conclusion: Existing evidence, although high level and concordant, has failed to convince doctors to uniformly embrace treatments for myopia progression control, possibly due to existence of some heterogeneity, reporting of side effects and lack of long-term follow-up. Research geared towards efficient interventions is still necessary.
OCT findings and visual prognosis in children with unilateral myelinated retinal nerve fibers and high myopia

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Introduction: This study focused on the visual prognosis of children with unilateral peripapillary myelinated retinal nerve fibers (MRNF) associated with myopic anisometropia as well as amblyopia, and to compare the macular OCT imaging characteristics of normal and myelinated eyes.

Methods: Seven children with unilateral MRNF were included, all children were corrected by contact lenses, given occlusion therapy, and were followed with good compliance. Macula OCT was performed with the Heidelberg Spectrals OCT ART 1 20x20º programme and the choroid was visualized with spectral domain enhanced-depth imaging optical coherence tomography (SD-EDI-OCT, Heidelberg Spectralis). The main outcome measures were visual acuity, central foveal thickness, perifoveal thickness in 8 surrounding quadrants named as EDTRS fields, and subfoveal choroidal thickness.

Results: Mean age at the initial visit was 3.88±2.95 years (range 1.2-8 years), mean follow-up time was 101.38±25.76 months (range 60-124months). Mean refractive error in spherical equivalents was -12.55±3.49 diopters (range -9.00 to -17.50 diopters). Visual acuity of the myelinated eye at the first visit was 0.68±0.39 logMAR (range 0.30 to 1.30 logMAR) which improved to 0.18±0.13 logMAR (range 0.30 -0 logMAR) at the last visit (p=0.04). Mean central retinal thickness (CRT) was 290.80±98.17µm in myelinated eyes and 249±23.33µm in normal eyes (p=0.40). Mean subfoveal choroidal thickness was 210.8±96.28 µm in myelinated eyes and 298.2±32.9 µm in normal eyes (p=0.10)

Conclusion: Occlusion therapy in patients with MRNF and high myopic anisometropia was successful in our patient group. CRT and CCT were not significantly different between the myelinated and normal eyes.

SD-OCT and FAF at onset of inherited maculopathy in children

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Introduction: In children, retinal and macular inherited dystrophies could be misdiagnosed at early stages when the symptoms are mild and there are no observable fundus changes. Our study of spectral domain OCT (SD-OCT) and FAF retinal imaging in the children with known familial history of maculopathy allowed to describe precocious abnormalities and to establish distinctive features in Stargardt disease (STGD), Juvenile Neuronal Ceroid Lipofuscinosis (JNCL or NCL3)and cone dystrophy (COD).

Methods: OCT and FAF imaging were analyzed in three children with a familial history of STGD, NCL3 and COD. SD-OCT was performed with Cirrus SD-OCT or Spectralis OCT 2, Heidelberg. FAF images were realized with Heidelberg Retina Angiograph.

Results: The most prominent common sign in the case of STGD and in NCL3 was a bulged, fluffy appearence of subfoveal external limiting membrane (ELM)at OCT. Though, the distinctive feature in STGD was local perifoveal interruption of ellipsoid line (EL) and in NCL3 a thinning of inner nuclear layer. Conversely, in COD there was no apparent changes of ELM. The earlier OCT finding in this entity was patchy interruption of perifoveolar EL.

The FAF images were distinctive in this three diseases. Little hyperautofluorescent perifoveolar dots were found in STGD. In NCL3 there was a hypoautofluorescent perifoveolar halo. Hyperautofluorescent perifoveolar ring was specifical in COD.

Conclusion: OCT and FAF are the first tests to be performed when the maculopathy is suspected in children. It could be enough to establish a diagnosis if the familial history is positive. OCT and FAF imaging could be useful in differential diagnosis of inherited maculopathies at earliest stages with a view to future retinal therapies.
Torpedo maculopathy: a multimodal analysis of a pediatric case series

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Introduction: Torpedo maculopathy (TM) has first been described in 1992 as a focal congenital abnormality of the retinal pigment epithelium (RPE) consisting in an asymptomatic torpedo-like shape lesion located close to and pointing the temporal portion of the macula. We describe a series of children affected by TM and characterize this lesion in a multimodal way.

Design: Retrospective observational case series.

Methods: Children examined in our clinic between 2014 and 2017 and having presented with TM were retrospectively collected. Visual acuity, orthoptic evaluation, fundoscopy, fluorescein and ICG angiography, OCT, fundus autofluorescence and automated visual fields were analyzed.

Results: Three children (2M:1F), i.e. 2 RE:1 LE, were included. Median age at the diagnosis was 50 months. Visual acuity was symmetric after refractive correction with no significant amblyopia. Intermittent convergent strabismus was seen in one affected eye. Fundoscopy of the two first cases revealed a classic, well-circumscribed torpedo-like chorioretinal lesion pointing the fovea with a slightly pigmented temporal tail, located in the horizontal raphe. The third case presented with the same clinical aspect, but the lesion was located in the retinal superior-temporal blood vessels. Fluorescein and ICG angiography showed transmission hyperfluorescence due to RPE atrophy and no abnormal leakage. No autofluorescence was detected, except in the margin of the TM. Subfoveal disruption of the photoreceptor and RPE complex with thinning of the outer retinal layers and hyperreflective choroid was always seen on OCT. However, cavitation defect was not always present. Automated visual field revealed a paracentral scotoma in one case. Toxoplasma serology was negative in one patient and maternal serology in the two others. Parental fundoscopy was physiologic in all cases.

Conclusion: TM seems not to be always located exactly in the horizontal raphe and could be slightly upper deviated. Differential diagnosis includes torpedo-like lesion along the arcades associated with enhanced S-cone syndrome (ESCS) or with toxoplasma chorioretinitis. However, our patient had negative toxoplasma serology and no other retinal features compatible with ESCS. Furthermore, the two types of OCT presentation could be linked to different evolution stages with cavitation cleft occurring later in life as in Best disease.

Validity and usability of new stimuli for colour vision assessment in infancy

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Introduction: Although colour blindness affects around 8-10% of infants, colours are frequently used in educational resources. However, there is a lack of standardized quantitative tests for colour perception in children younger than 3 years of age. The purpose of this study was to evaluate validity and usability of a novel test for assessing colour vision perception in children from 1 to 24 months of age.

Methods: A total of 57 children participated in this study, run in the BabyLab, Department of Experimental Psychology, University of Oxford. Our colour test uses pseudoisochromatic plates displayed on a monitor. Nine chromaticity pairs along the three theoretically significant lines in colour space (the protan, deutan and tritan confusion lines) were presented. The study comprised two sequences of 9 visual stimuli each, with a total duration lower than 2 minutes. Colour perception was assessed based on their visual behaviour. Eye movements were tracked using a Tobii TX300 Eye Tracker (Tobii Technology AB, Sweden).

Results: Of the 51 children older than 3 months of age, 43 underwent the complete test, 7 completed the first sequence only and 1 had to be excluded because of lack of attention to the visual stimuli. Although immature, colour vision can be quantified in three-month-old children. It improves incrementally during the first months of life on all the three colour axes. As in adults, colour discrimination in deutan and protan axes seem to be more accurate than in tritan in young infants.

Conclusion: We propose a novel objective and quantitative test for assessing colour perception in children older than 3 months.
**Assistive technology for children with low vision: the CREATE pilot trial (Children Reading with Electronic Assistance To Educate)**

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**Introduction:** Low vision adversely affects education and independence of children and young people. New technologies such as tablet computers can provide assistance for accessing information and processing text by enlarged font, speech output and voice recognition. The aim of this pilot trial was to test the methodology for a full randomized controlled trial (RCT) which will assess the impact of tablet computers on education and access to information in students with low vision.

**Methods:** We randomized 40 Students in the UK and India, aged 10-18 years and with low vision, to either Apple iPads (active intervention) or local standard low-vision aid care (control). At baseline and at 3 and 6 months, we recorded trial feasibility measures (recruitment, loss to follow-up) and acceptance/usage and accessibility of the device (primary outcomes) and assessed vision-related secondary outcomes: vision-related quality of life, validated measures of reading and comprehension, and adverse effects.

**Results:** Despite a high volume of patients in low vision clinics, recruitment in the UK was more successful when led by the teachers from the Visual Impairment team. Two students were withdrawn from the control arm, as schools provided tablet computers during the course of the study. One participant in the iPad group was withdrawn, as the device was not supported by teacher. Preliminary analysis of secondary outcomes did not demonstrate significant change.

**Conclusion:** This is the first RCT of a low-vision technology for children; pilot data demonstrate that a full RCT would be feasible. Students, parents and most teachers commented favourably on the usefulness of tablet computers for learning and accessing information independently.
Inherited retinal dystrophies are a group of diseases that may manifest in all periods of life. First symptoms most commonly occur in childhood or adolescence. Their unifying features include their underlying genetic aetiology and the progressive degeneration of photoreceptors. Their pathology may be limited to the eye or it may be part of a syndrome, such as a ciliopathy, mitochondriopathy, or a metabolic disease. Clinically, differentiation of retinal dystrophies largely depends on electoretinography, although a molecular genetic classification is increasingly being used. Possible differential diagnoses that may mimic retinal dystrophies include stationary inherited retinopathies, post-inflammatory, toxic, or autoimmune disorders.

With today’s routinely used imaging modalities, retinal alterations can be detected in virtually all symptomatic patients with retinal dystrophies. Moreover, novel methods such as wide field fundus autofluorescence imaging, spectral-domain OCT, adaptive optics and quantitative autofluorescence may even allow pre-symptomatic detection of disease. Such “predictive imaging” may have substantial psychological and legal implications and should be performed in conjunction with genetic counselling.

Overall, an accurate diagnosis can only be made based on comprehensive assessment of the disease manifestations. This will guide counselling the patient about the expected disease course and inheritance, to encourage low vision rehabilitation, and to interpret results of genetic testing. Lastly, high-quality phenotyping (and genotyping) is necessary to provide information on current and future treatment trials that may slow the progression of photoreceptor loss or restore vision.
Electrodiagnostic testing for inherited retinal dystrophies

Dorothy Thompson

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We can access exquisite images of retinal structure yet these give us little idea of function. To find this out we need objective visual electrophysiological tests. These electrodiagnostic tests can identify and characterise a retinal dystrophy. They include three types of electroretinogram (ERG), which are described by the stimuli used; e.g. flash (fERG), pattern (PERG) or multifocal (mfERG). Structured patterns localise the area of retina assessed. There is also the electro-oculogram (EOG) and the visual evoked potential (VEP), to both flashes and patterns.

Standard protocols published by the International Society for Clinical Electrophysiology of Vision (ISCEV) ensure the same result may be obtained anywhere in the world. The ISCEV ERG tests demand 20 minutes dark adaptation and pupillary dilation, which means that pattern ERG and VEPs must be performed at a separate session to the flash ERG. For young children the ISCEV flash ERG protocol can be achieved by using anaesthesia, but this can reduce and delay the ERG. At GOSH we have developed an alternative protocol that produces physiological comparable data in alert children, in a shorter time. Typically within the same 30 minute test session a skin flash ERG is carried out with a pattern VEP that provides an index of macular pathway function.

Flash ERGs distinguish generalised cone and rod, outer and inner, retinal function. A few conditions, e.g. KCNV2 and eScone syndrome, have constellations of ERG that are pathognomic. The pattern ERG distinguishes maculopathies from ganglion cell and optic nerve dysfunction. An EOG indicates the functional relationship between RPE and photoreceptor cells and has been used more often since the wider phenotype of AR bestrophinopathies has been elaborated. All results are a snapshot in time; they may monitor progression or treatment effectiveness e.g. IAM for retinoblastoma, but require knowledge of the fullest clinical context for the best interpretation. Case examples highlighting these applications will be discussed.

www.facebook.com/ISCEV.org
Inherited retinal degenerations (IRDs) are a leading cause of blindness and visual loss. IRDs are considered as rare diseases (disorders that affect less than 5 in 10,000 in the population). For a significant proportion of rare diseases, the cause can be traced to mutations in a single gene, referred to as monogenic, typically associated with Mendelian inheritance. Around 80% of rare diseases are currently thought to be genetic. Over the last 30 years there has been a rapid evolution in our ability to identify disease causing genes. NGS, introduced in the mid 2000’s has enabled the rapid sequencing of large amounts of DNA, changing the face of genetic testing since the first gene was mapped in 1983. Gene panels, whole exome and whole genome testing together with other genetic testing strategies have taken this to a new level. IRDs are genetically extremely heterogeneous with over 250 genes identified thus far. In children with IRDs the diagnostic testing yield using NGS can reach 85%. Reasons to carry out genetic testing include diagnosis, prognosis, clinical management, genetic counselling, and assessing eligibility for gene therapy trials. There is still significant complexity in genetic testing: finding pathogenic variants in more than one gene- leading to the question as to which is the relevant gene; identifying only one variant in an autosomal recessive condition; tackling unsolved cases; refining bioinformatics pipelines and algorithms with sequential confirmation of pathogenicity of a variant; identifying non-coding disease causing variants, and evaluating the role of genetic modifiers are key examples.
Retinal gene therapy has proven to be safe and effective in some cases for treating rare single gene disorders causing retinal degeneration. Most advanced is the adeno-associated viral (AAV) vector treatment for Leber Congenital Amaurosis caused by recessive RPE65 mutations in the programme led by Spark Therapeutics. Recent progress in choroideremia gene therapy has been made by the University of Oxford in partnership with Nightstar Ltd. (Nightstar), which has led to a multicentre international Phase II and forthcoming Phase III clinical trials, spanning 8 different countries. A scientific approach of using codon optimisation to stabilise the RPGR transgene has led to the first clinical trial using gene therapy to treat X-linked retinitis pigmentosa - the most common recessive variant of the disease - in a Nightstar sponsored Phase I/II clinical trial currently ongoing in Oxford and Manchester in the UK. A brief overview of the science behind these developments will be presented.
How to explain “flat” electroretinograms when patients with Leber’s congenital amaurosis aren’t blind

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Introduction: To explain how a “flat” or “extinguished” electroretinogram (ERG) in a patient with Leber’s congenital amaurosis can be diagnostic for this entity, yet not necessarily be indicative of severe loss of vision.

Methods: Basic principles of ocular as well as auditory electrophysiology, wave physics, phototransduction metabolic pathways and ophthalmic genetics were reviewed and organized to reveal heretofore overlooked pathways. Retrospective observation and correlations of ocular as well as auditory electrophysiology characteristics for Leber’s congenital amaurosis and congenital auditory neuropathy/dys-synchrony were made. The literature was searched using MEDLINE, and articles were obtained from the bibliographies of these publications.

Results: Variably delayed individual photoreceptor cell signaling secondary to metabolic enzymatic deficiencies involved in phototransduction results in ERG a-waves produced, and subsequent b-waves, to interfere destructively with each other, rather than allow for averaging in-phase summation to permit recordable ERG signals. Despite the delayed visual cycle, intraretinal synaptic transmission may nonetheless proceed uninhibited to activate retinal ganglion cells and axonal pathways to the visual cortex.

Conclusion: A “flat” or “extinguished” ERG need not indicate absent vision, but overall destructive interference of individual a-waves and b-waves averaged for the recorded ERG. Analogous auditory electrophysiologic findings are noted in corresponding so-called congenital auditory neuropathy/dys-synchrony. Such findings are of great diagnostic utility and may be considered pathognomonic. Review of basic principles and the literature indicate, moreover, that previously investigated, but since neglected, electrooculography may offer benefits in terms of prognostic value. Theoretically, the stability or progression of a patient with Leber’s congenital amaurosis, and their response to an interventional genetic or pharmacologic therapeutic regimen, could be followed in more sensitive and objective manner at the cellular level via electro-oculography, prior to visual acuity, visual evoked potentials, or ERG testing revealing any changes in overall retinal function.
Genotype-Phenotype Correlation in inherited retinal disease

CEP290 variations

Introduction: Inherited retinal disease (IRD) are a major cause of visual impairment in children. Recent advances in high throughput DNA sequencing technologies have revolutionised genetic testing approaches. The aim of this study is to assess the current clinical validity and utility of genetic testing in children with IRD.

Methods: 86 unrelated children (≤16 years old) with a diagnosis of IRD were ascertained through tertiary ophthalmic genetics clinics at Central Manchester University Hospitals, Manchester UK. Study subjects underwent a detailed clinical assessment, electrodiagnostic testing (EDT) in 72 participants, where appropriate. The diagnostic yield of clinical genetic testing was subsequently performed in an accredited diagnostic laboratory. Only individuals presenting between January 2014 and July 2016 were included.

Results: Overall, 7.6% of patients presented with a molecular diagnosis in paediatric IRD. The genes most frequently mutated in these study subjects had a spectrum of clinical features consistent with syndromic IRD subtypes). The most frequent variation was p.K1557* (10 / 48 alleles) followed by the frequent deep intronic variation c.2991+1655A>G (5 / 48 alleles). The second frequent variation was present in 20 patients (27 / 48 alleles), among these 7 patients were homozygous for the variation. The second frequent variation was c.2119_2123dupCAGCT (5 / 48 alleles). Amaurosis was noted in 7 patients during the first months of life. All patients followed over the second year of life developed at least light perception (LP, n=7) or hand movement (HM, n=8). In 8 patients BCVA could be quantified (0.008 - 0.2) even up to the age of 77. All patients had electrophysiological responses below threshold.

Conclusion: Preserved ganglion cell and nerve fiber cell layers may provide a target for upcoming therapeutic approaches.

The Diagnostic Yield of Clinical Genetic Testing in Pediatric Inherited Retinal Disease

Methods: One hundred and seventy-six patients with IRDs were ascertained through tertiary ophthalmic genetics clinics at Central Manchester University Hospitals, Manchester UK. Study subjects underwent a detailed clinical assessment, including distinguishing stationary from progressive and isolated phenotypes. Study subjects underwent a detailed clinical assessment, electrodiagnostic testing and photomicroscopy. The diagnostic yield of clinical genetic testing was subsequently performed in an accredited diagnostic laboratory. Only individuals presenting between January 2014 and July 2016 were included.

Results: Overall, 53% of patients presented with a molecular diagnosis in paediatric IRD. The genes most frequently mutated in these study subjects had a spectrum of clinical features consistent with syndromic IRD subtypes). The most frequent variation was p.K1557* (10 / 48 alleles) followed by the frequent deep intronic variation c.2991+1655A>G (5 / 48 alleles). The second frequent variation was present in 20 patients (27 / 48 alleles), among these 7 patients were homozygous for the variation. The second frequent variation was c.2119_2123dupCAGCT (5 / 48 alleles).

Conclusion: Panel-based clinical genetic testing pointed to a molecular diagnosis in 84.7% of children with IRD. The diagnostic yield in the paediatric population was found to be significantly higher compared to that in adult IRD cohorts (84.7% vs 5%). Approaches similar to the one described here are expected to become a standard component of care in pediatric IRD, accelerating diagnosis and facilitating precision medicine approaches.

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Conclusion: Preserved ganglion cell and nerve fiber cell layers may provide a target for upcoming therapeutic approaches.
10 year audit of The Paediatric Low Vision Aid Clinic at Oxford Eye Hospital

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Introduction: The Paediatric Low Vision Aid Clinic forms part of a multidisciplinary service supporting children and young people (C&YP) with visual impairment across secondary care, in their community and education. This presentation gives 10 year data from the clinic including diagnosis, level of visual impairment and ongoing management outcomes.

Methods: A secured clinical database was analysed using standard statistical methods, exploring cross sectional and longitudinal data.

Results: A total of 216 patients have been seen in the clinic since 2003.

In 2017 there were 108 C&YP under active review; 59 male, 49 female. Average age at presentation was 6 years. Average visual acuity (VA) at presentation was 0.655 LogMAR. Leading causes of visual impairment was nystagmus (n = 39) followed by Retinal Dystrophy (n = 26).

Longitudinal data is presented for differing pathologies looking at both change in VA and change in magnifier use over time.

Conclusion: This is, to the author’s knowledge, the first time that 10 year data has been presented. The results will be used in planning future service provision and development.

Combined nonsense-mediated decay inhibition and translational readthrough as a treatment approach for hereditary retinal dystrophies

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Introduction: Nonsense-mediated decay (NMD) is a protective evolutionarily preserved cellular machinery which degrades mRNA transcripts containing nonsense mutations. As translational bypass therapy is becoming a reality for the treatment of nonsense-mediated disease, it may be counter-productive to degrade the substrate which has potential to be translated into functional protein. Choroideremia (incidence 1:50,000) is an X-linked recessive, progressive, chorioretinal dystrophy with a third of cases harbouring nonsense mutations in the CHM gene encoding REP1 protein. Amlexanox is a well-established anti-inflammatory drug with the combined action of NMD inhibition and translational readthrough, which may be an effective treatment for nonsense-mediated retinal dystrophies.

Methods: We tested the safety and efficacy of amlexanox for the treatment of nonsense-mediated choroideremia using patient fibroblasts and the chm-/- zebrafish model. Dose-response experiments were conducted to determine the safest and most efficacious dose of amlexanox in the chm-/- zebrafish (Q32X, UAA stop) and CHM patient fibroblasts (Y42X, UAG stop). 100 µM was administered to zebrafish at 8 hours post-fertilisation and 300 µM to fibroblasts. Survival studies, histology, cell death assays, qRT-PCR, western blot and functional prenylation assays were undertaken (n=3 independent experiments).

Results: Mean survival (± SEM in days) of amlexanox-treated chm-/- zebrafish was 9.4 ± 2.1 versus untreated zebrafish 4.8 ± 1.0 (p<0.01, n= 70 per group). At day 6, there was no evidence of retinal degeneration in treated mutants; qRT-PCR showed a 1.8-fold increase in mRNA transcripts indicating NMD inhibition, 50.6% increase in REP1 and 66% decrease in unprenylated Rabs, indicating significant rescue of functional REP1 protein. Treated CHM fibroblasts showed a 1.1-fold increase in mRNA and 18.8% decrease in unprenylated Rabs.

Conclusion: This study provides evidence for amlexanox as an effective NMD inhibitor and readthrough drug for treating nonsense-mediated choroideremia. NMD inhibition may provide a vital new treatment strategy for other hereditary retinal dystrophies, which may previously have been untreatable.
Post infectious pigmentary retinopathies in infants

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Introduction: The dissociation between anatomy (a pigmentary retinopathy on fundoscopy) and function (a retinal dystrophy, which means a progressing retinal dysfunction on global ERG) is a challenging situation for ophthalmologists, especially in paediatrics.

Methods: Four cases of infants presenting with a clinical picture of pigmentary retinopathy and either initially normal ERG, or a stable, post-infectious retinopathy, are reported.

Results: In two cases, other features allowed to diagnose a foetal infection. In one case, a severe and acute retinopathy with features of a retinal dystrophy, except for the timing - acute and stable - occurred after a varicella infection. In the last case, a severe pigmentary retinopathy was associated with an encephalopathy; global ERG was initially normal (foetal infectious sequellae were initially hypothesised), before slowly progressing and allowing to diagnose a retinal dystrophy.

Conclusion: Dissociation of structure and function in infantile retinal disorders may preclude any diagnosis at presentation. Multimodal imaging and repeated electrophysiological assessments are here necessary for the diagnosis.

Choroidal neovascularization secondary to Best vitelliform macular dystrophy: is it rare?

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Introduction: Best vitelliform macular dystrophy (VMD) also known as best disease is an autosomal dominant retinal dystrophy caused by heterozygous mutations in the bestrophin 1 gene. Histopathologic evaluations, Spectral domain optical coherence tomography and fundus autofluorescence indicated lipofuscin accumulations at the level of the retinal pigment epithelium (RPE) and also revealed atrophy and disruption of the RPE/photoreceptor complex. In rare cases were reported VMD is accompanied by formation of a choroidal neovascular membrane (CNVM) that is most often observed at a scarring stage. Single cases of development of CNVM at vitelliform stage are describe.

Purpose: To report the optical coherence tomography angiography (OCTA) features of CNVM of VMD.

Methods: Retrospective serial case reports of patients with VMD who underwent OCTA of the posterior pole.

Results: We report a 6 cases of CNVM secondary to Best VMD in 10 eyes of 4 children aged 5-12 and woman aged 25 that was detected using OCTA. VA was 20/25-20/20. In all cases CNVM was detected in zone under accumulation of lipofuscin substance at vitelliform and pseudohypopyon stages of VMD.

Conclusion: OCTA is a non-invasive, effective imaging technique that can offer additional information about the vascular characteristics of macular lesions in various pathologies. Our study demonstrates the capability of OCT-A to allow early diagnosis of the presence of a CNVM in VMD patients and leaves an issue about the true frequency of its development is open.
Intravitreal anti-VEGF treatment for choroidal neovascularisation secondary to best vitelliform macular dystrophy in a 5-year-old child and his grandmother

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Introduction: To report about a child and his grandmother with choroidal neovascularisation (CNV) secondary to Best vitelliform macular dystrophy who recovered good visual acuity with intravitreal antiVEGF treatment.

Methods: Nine patients of the same Slovene family with Best vitelliform dystrophy with confirmed VMD2 mutation and abnormal EOG will be presented. Two of them were treated with antiVEGF due to secondary choroidal neovascularisation - the grandmother and her four years old grandson. The boy was referred to us due to reduced vision bilaterally seen on a routine vision screening exam at the age of four years. He was hypermetropic, with glasses his vision improved to (RE:0.6cc, LE:0.3cc; Snellen). On fundoscopy there were bilateral vitelliform lesions seen centrally in the maculae, that were hyperautofluorescent on autofluorescence imaging.

Results: 6 months later his vision in the left eye dropped to 0.2cc, the haemorrhage was seen centrally at his fundus and lesion was suspected for choroidal neovascularisation (CNV). Fluorescein angiography and OCT were done, they showed classic choroidal neovascularisation in his left eye. He was treated with bevacizumab intravitreally left eye (twice). Later his vision dropped to 0.1cc in his right eye, OCT and fluorescein angiography showed classic choroidal neovascularisation in his right eye. He was treated with bevacizumab intravitreally right eye (three times). In the period of nine months after antiVEGF treatment his vision improved bilaterally (RE:0.4cc, LE:0.6cc), new glasses were prescribed and occlusion of the better left eye was recommended. OCT and angiography demonstrated regression of the CNV and resolution of macular edema. At his last visit after four years his vision improved bilaterally (RE:0.8cc, LE:0.8cc).

Conclusion: Intravitreal anti-VEGF treatment may be effective resulting in morphologic and functional improvement in children suffering from CNV secondary to Best vitelliform dystrophy. Only a few injections of antiVEGF with additional orthoptic treatment and regular follow up were needed in our patient.
**Hereditary retinal diseases diagnosed in infants with congenital vertical nystagmus**

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**Introduction:** In the present study, we describe retinal aetiologies of congenital vertical nystagmus (CVN). The main goal was to establish a strategy of exams in the setting of CVN.

**Methods:** Our retrospective cohort included all paediatric patients with diagnosis of CVN. All patients underwent complete ophthalmic examination with nystagmus movie, visual electrophysiology testing (VEP+ffERG), neuropediatric examination and brain MRI.

**Results:** We recruited 48 patients with CVN from 2006 to 2017. At the basis of nystagmus movie analysis, 35% of infants had an up-beat nystagmus, 30% had a down-beat nystagmus, 25% had a pendular vertical nystagmus and 10% had a vertico-rotatory nystagmus. Overall, 40% of patients have been diagnosed with anterior visual pathway disease. The main “ophthalmic” aetiologies of CVN were: stationary photoreceptor dysfunction syndromes (achromatopsia, blue cone monochromacy, congenital stationary night blindness), early retinal degenerations (non-syndromic Leber amaurosis and Joubert syndrome) and albinism. 38% CVN were “neurologic” in origin. 2 cases of optic nerve hypoplasia et 2 cases of optic nerve gliomas were discovered. 16% were diagnosed as idiopathic.

**Discussion:** Anterior visual pathway diseases were at least as frequent as neurological conditions in patients with CVN. In our cohort, stationary retinal dysfunctions and early onset retinal degenerations were prominent “ophthalmic” aetiology of CVN.

**Conclusion:** We believe that a complete ophthalmic examination with a visual electrophysiology testing (VEP+ffERG) combined with neuropediatric examination and a brain MRI should be realised in all infants with CVN.
The maintenance of oxygen homeostasis is a fundamental physiological challenge requiring precise co-ordination of oxygen delivery with metabolic requirements. Nevertheless for most of the twentieth century, efforts to understand the process focused mainly on the metabolic consequences of hypoxia rather than direct sensing of molecular oxygen. That paradigm was overturned by studies of cis-acting control sequences at the erythropoietin gene locus, which led to the recognition of a general system of direct oxygen sensing in animal cells that controls genes involved in energy metabolism, angiogenesis, erythropoiesis, cell differentiation and numerous other cellular or systemic responses to hypoxia. The process is mediated by a series of 2-oxoglutarate dependent dioxygenases that catalyse the post-translational hydroxylation of specific amino residues in the transcriptional mediator, hypoxia inducible factor (HIF). The lecture review the elucidation of these pathways, and consider their potential therapeutic tractability.
Acute retinopathy of prematurity ROP is the result of premature birth and subsequent abnormal vascular development due to activation of the hypoxia induced factor HIF1 leading to significant changes in growth factors such as vascular endothelial growth factor VEGF and insulin growth factor IGF1. Despite significant advances in neonatal care and knowledge acute ROP is still a potentially blinding disease in developed countries as extremely premature infants with extremely immature vascular system at birth are surviving, and in developing countries due to shortage of staff and instrumentation. The clinical presentation depends on the degree of prematurity and prenatal, perinatal and postnatal problems. An international classification describes the various degrees of extent and severity of ROP. Multiple studies have defined the forms needing treatment with the Early Treatment of Retinopathy of Prematurity ETROP recommendations being the most widely accepted an at present followed ones. Nation specific guidelines have been developed to detect infants needing treatment in due time. Classification may show subjective bias as evident from studies comparing expert decisions based on wide-field retinal images. Retinal wide-field imaging including telemedicine has indeed the potential to objectively compare inter observer classifications, and questions binocular indirect ophthalmoscopy BIO as the gold standard. Fluorescein angiography FA has opened a new field of evaluating the extent of vascularisation including avascular zones central to the peripheral vascularisation border. In addition, FA visualizes the degree of vascular leakage which is particularly intense in aggressive posterior ROP APROP. Hand-held spectral-domain optical coherence tomography hh-SD-OCT discloses knew features such as macular oedema and macular immaturity that may later translate into macular developmental arrest MDA responsible in part for reduced visual function despite regression of acute ROP. This paper will focus on the newer aspects of diagnosing and classifying acute ROP.
Clinical and surgical management of ROP

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The lecture on clinical and surgical management will briefly cover standards of care followed by presentation of more controversial issues including use of anti-VEGF agents and Intraocular surgery for tractional retinal detachment.
Findings from the national UK Retinopathy of Prematurity treatment study

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Introduction: At EPOS 2016 we reported that between December 2013 and December 2014 327 infants born preterm in the UK required treatment for ROP. Here we present further details on severity of ROP leading to the decision to treat, re-treatment rates and anatomical and refractive outcomes at one year.

Methods: National surveillance study supported by the British Ophthalmic Surveillance Unit (BOSU). Clinicians completed an initial form (demographic and clinical data about primary treatment and early re-treatment), and 12 months later a second form with further re-treatment, anatomical and refractive details.

Results: 204 right eyes (RE, 62.39%) had type 1 ROP, and 27 (8.26%) had aggressive posterior ROP. Infants were also treated for milder disease: 9 (2.75%) RE were treated for type 2 ROP; and 74 (22.63%) for disease milder than type 1 with plus or pre-plus, which we defined as “type 2 plus” disease. First-line treatment was diode laser photoablation in 90.5% (297 RE) and injection of vascular endothelial growth factor inhibitor (VEGF-I) in 8% (26 RE). Follow-up data on whether re-treatment was required were available for 186 infants (57% of the original cohort; 169 with both eyes treated and early re-treatment), and 12 months later a second form with further re-treatment, anatomical and refractive details.

At one year, the macula of two RE (1.4%) was “dragged”, and one RE (0.7%) had a retinal fold involving the macula. Best corrected visual acuity in logMAR in the RE was available in 46 children, and with both eyes open (BEO), 89 children. Median (IQR) RE acuity was 0.6 (0.4 to 1.0), and median acuity BEO 0.4 (0.3 to 0.7) logMAR. Median spherical equivalent of RE was 0.44 (-1.3 to 1.3) dioptre (n=116). Median astigmatism of RE was 0.5 (0 to 1.0) dioptre (n=111). Median anisometropia was 0.125 (0 to 0.75) dioptre (n=116). 24 children (20.5%) had been prescribed glasses. Eleven (8.4%) children had been certified as sight impaired, and five (3.82%) as severely sight impaired.

Conclusion: A new category, “type 2 plus” ROP, lead to a decision to treat in a significant number of infants. VEGF inhibitors are used in a small proportion of cases. Re-treatment rates after VEGF-I are higher than after primary laser treatment.
Is ROP the worst abnormality what we can find in a preterm baby?

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Introduction: According to the international guidelines the initial eye examination is suggested on the 30th-31st postmenstrual week for babies born before the 27th week of gestation and on the 4th postnatal week for more mature children who are at risk for development of ROP. Early detection of other eye abnormalities can be possible.

Purpose: to show different ocular pathologies with and without known etiology in preterm children screened in Semmelweis University or referred to our department in the last 10 years.

Methods: Our retrospective analysis included data of 27 children (16 girls, 11 boys). Mean gestational age was 29.07 weeks (24-35; SD: ±2.69) at birth and mean birth weight was 1048.2 (650-1980; SD: ± 334.1) grams.

Results: Abnormalities of anterior and posterior segment were recognised: anophthalmos (1), PFV (1), dislocated lens (10), abnormal optic nerve head (3), macular haemorrhages without ROP (2), vascular abnormalities (1), RPE changes (3) and choroidal lesions (7).

Conclusion: More data and further analysis are needed to answer what is the cause of these findings and how can we protect the babies from them.

Prediction of ophthalmological problems in 6.5 year-old prematurely born children - preliminary results of a population-based, prospective study

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Introduction: Ophthalmological and visual problems are common in prematurely born children, but there is no consensus on the long-term ophthalmological follow-up.

Methods: The aim of the study was to describe the ophthalmological outcome at 6.5 years and to identify possible predictive factors visual problems at this age.

Results: Mean visual acuity (VA) did not differ between the preterm and full-term children, but a "subnormal" VA of ≤ 0.8 was more common in the preterm group (31% versus 14%) (p<0.05). No prematurely born child was blind, but two had VA < 0.5. A contrast sensitivity of < 0.5 was more common (36% versus 19%, p<0.05) and strabismus was also more common in the pre-terms (8% versus 0%, p<0.05) than in the controls. Within the preterm group, logistic regression analyses revealed GA, ROP, PVL/IVH, and mild cognitive disability (Bayley III test), to be predictive factors for ophthalmological and visual problems at 6.5 years age.

Conclusion: Prematurely born children had an increased prevalence of ophthalmological problems at 6.5 years age. Treated ROP and early neurological problems were risk factors for later ophthalmological dysfunctions. Further, this study showed that reduced cognitive function at 2.5 years appeared to predict such problems.
The macula zone status in cicatrical period of retinopathy of prematurity according to optical coherence tomography

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Introduction: For the vision formation in children in the cicatrical period of retinopathy of prematurity (ROP), the features of the anatomical structure of the macular area are of great importance: primary, due to incomplete retinal embryogenesis by the time of preterm delivery and secondary, as a result of the vasoproliferative process in acute ROP. Optical coherence tomography (OCT) allows non-invasive, with high accuracy and reliability to investigate the central parts of the retina.

Purpose: To study the status of macula zone of the retina in the cicatrical period of retinopathy of prematurity (ROP) according to optical coherence tomography.

Material and Methods: Data of visometry, refractometry and axial length were evaluated in 90 children (174 eyes) in the cicatrical ROP period. The gestational age in the examined patients ranged from 24 to 36 weeks, the body weight at birth was 460-2400 grams. The age of children was from 3 years 4 months to 17 years 6 months (11.24 ± 0.86 years on average), boys - 40 (44.4%), girls - 50 (55.6%). In addition to standard ophthalmological examinations in 84 children (93.3%) OCT of the macula zone was performed. Due to the residual central fundus changes all the patients were divided into 3 groups. The control group - full-term children with myopic refraction (31 persons, 62 eyes), comparable in age and sex.

Results: A reduction or an absence of foveolar depression is typical for the OCT of the macula in the cicatrical ROP period with a maintence of the triangular sideview of the outer nuclear layer in fovea centralis projection. A decrease of the neuroepithelium volume in the macula with simultaneous increase of the neuroepithelium thickness in the fovea was revealed, indicating a hypoplasia of the macular area in the premature children in the cicatrical period of the disease. The discontinuity or the rarefaction of the junction line of the outer and inner segments of photoreceptors and the pigment epithelium complex-choriocapillaries were noted in a maintence of the normal structure of layers in all groups of observation due to incomplete differentiation of the fovea microstructure in preterm patients. The OCT data correlated with the data of visometry and refractometry in premature infants in the cicatrical period of the disease: the more significant OCT changes were revealed, the less visual acuity was rated. Even minimal OCT changes do not allow ROP patients to get maximal visual acuity.

Conclusion: The OCT of the macular area is a highly informative non-invasive method in diagnosis of the cicatrical ROP.

Late retinal detachment in adult retinopathy of prematurity

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Introduction: Patients with favorable outcomes of retinopathy of prematurity (ROP) are at the high risk group for various vitreoretinal pathology, which is based on abnormal vitreoretinal traction and disturbance of the vitreoretinal interface, changes in the course of peripheral and central vessels, ruptures and retinal detachments (RD) in children and adults with regressive ROP. According to foreign authors, the frequency of late RD is 14-26% , according to Russian data, 12%.

Materials and Methods: In 2014-2016 126 patients (246 eyes) with a favorable outcome in the cicatrical period of ROP were examined. The age of children was from 3 years 4 months to 17 years 6 months (11.24 ± 0.86 years on average), boys - 58 (46%), girls - 68 (54%).

The gestational age in the examined patients ranged from 24 to 36 weeks, the body weight at birth was 460-2400 grams. The observation group included patients with spontaneous regression (98 patients, 191 eyes) and with regression after surgical treatment during the active period (28 patients, 55 eyes).

Results: In 12 children a late retinal detachment was detected, 4 of them had a bilateral detachment, thus the frequency of late RD with favorable outcomes of ROP was 9.5%. The age of RD origin was 6 years-16.6 years, on average 11.6 years, 5.5-16 years after the acute ROP. In 14 eyes (10 children), the RD was spontaneous, in 1 eye-traumatic, 1 eye-postuveitis. Regmatogenic RD-7 eyes (43.8%), holes not found in 8 eyes (50%), 1 eye-exudative. Total RD-2 eyes (12.5%), subtotal-4 (25.0%), partial local - 10 (62.5%). All children were intellectually preserved, the common was the absence of typical RD complaints.

Conclusion: All patients with ROP history are at risk of late retinal detachments developing. According to our data, the frequency of late RD with favorable outcomes is 9.5%.

The preservation of the central parts of the fundus and high visual functions do not always correlate with the state of peripheral parts: dystrophies, ruptures and retinal detachments.

Patients with favorable outcomes of ROP should be trained in self-monitoring of vision quality, emphasize the need for timely ophthalmological examination in any deterioration of visual functions.

Prematurely born children need lifelong adequate dispensary supervision. Timely detection and treatment of vitreoretinal complications avoids the development of late RD.

Surgical treatment of RD in the cicatrical period of ROP is most often combined.
**Autosomal dominant neovascular inflammatory vitreoretinopathy**

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**Introduction:** The purpose of our study was to review the course of illness 7 members of affected family with autosomal dominant neovascular inflammatory vitreoretinopathy (ADNIV). ADNIV is a very rare, autoimmune disorder belonging to inherited vitreoretinal dystrophies, without systemic features. ADNIV is caused by the mutations in the CAPN5 gene. The earliest signs are vitreous cells and mild peripheral retinal ischemia. Later it is characterized by panocular inflammation, pigmented retinal degeneration, retinal and iris neovascularization, neovascular glaucoma, vitreous hemorrhage, intraocular fibrosis and membrane formation and tractional retinal detachment. There is reduced b-wave amplitudes in ERG testing.

**Methods:** 7 of 20 members of a four-generation family affected by ADNIV were followed up. They underwent a complete ophthalmologic evaluation. Visual acuity, slit lamp biomicroscopy and fundus were monitored. The image of macula was observed using the OCT. The fundus photographs were recorded. They were referred for genetic evaluation and ERG testing.

**Results:** In all patients we observed significant deterioration of vision with age. Presenile cataract, posterior uveitis with vitreous cells, papilledema, cystoid macular edema, preretinal membrane, raised of intraocular pressure were observed. The ERG testing showed the presence of ADNIV. The mutation in exon 6 CAPN5 gene was not found but this condition doesn’t exclude the presence of VRNI because there are many others mutations of CAPN6 gene which have not been available.

**Conclusion:** VRNI is a very serious progressing disease leading to visual loss. It is the one inflammatory vitreoretinopathy. The implementation of anti-inflammatory treatment is sometimes necessary in VRNI disease. Due to unavailability of genetic molecular tests the diagnostic process is prolonged and is based on amnestic, genealogy and clinical features with follow up.

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**Retinal injuries resulting from handheld laser devices in the paediatric population, A case series of 27 eyes**

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**Introduction:** Cases of retinal injury in the paediatric population resulting from the inappropriate use of handheld lasers has been reported with increasing frequency. Two published cases series have described macular injuries in paediatric patients and have reported cases with prolonged if not permanent reduction in visual acuity. We describe a case series collected in Northern Ireland of paediatric retinal injuries resulting from handheld laser exposure.

**Methods:** Review of suspected retinal laser injury cases presenting with evidence of visual loss and/or characteristic retinal findings. All included patients described exposure to a handheld laser and data on visual functional and retinal imaging was available for all cases. We also collected data on date of presentation, the laser source and any complications from the laser injury.

**Results:** 26 eyes/23 cases of retinal laser injury were identified presenting between Nov 2012 and Apr 2017. 2 patients were female with 21 males and the average age was 12 years old (Range 7-16 years). Mean visual acuity at presentation was 0.20 logMAR (6/9.5). Visual acuity was worse than 0.5 logMAR (6/12) in 6 cases at presentation with all but 2 of these improving to 0.5 logMAR (6/12) or better. OCT images were graded as per classification criteria proposed by Raoof 2016, with 12 “mild”, 8 “moderate” and 1 “severe” case.

Prior to a diagnosis 2 cases were initially investigated as possible infective retinitis following recent foreign travel. 1 further case presented with choroidal neovascularisation secondary to a retinal laser burn. The most common source of handheld lasers were those bought while on holiday outside the UK or Ireland i.e. 9 (39%). Referral and presentation of cases seemed to follow a seasonality pattern with a cluster of 14 (61%) cases presenting between September and November.

**Discussion/Conclusion:** Most cases had mild visual impairment with only 2 cases failing to improve to 0.5 logMAR (6/12). With regard to injury severity our findings agree those of Raoof et al. 2016, with better visual acuity correlating with a mild injury. We were however unable to adequately grade 1 case of choroidal neovascularisation with these criteria.

Many cases presented as incidental findings with a seasonal preponderance for the autumn months. We hypothesis this may relate to patients acquiring lasers while on summer break/holiday or as they attend an optometrist prior to the new school year.
Severe ocular complications in patients with macrocephaly-capillary malformation syndrome

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Introduction: Macrocephaly-capillary malformation syndrome (M-CM) is a rare, sporadic disease affecting the skin, vasculature and other organ systems and is part of the PIK3CA related overgrowth spectrum (PROS). M-CM can be associated with anterior or posterior segment ocular complications but no systematic analysis of this patient cohort is available. We present a case series of affected patients where 2 of them showed severe and partially unusual ocular complications.

Methods: Patients with M-CM received a detailed ophthalmological examination after the clinical diagnosis was established by the pediatric dermatologists. One of the patients received additional ROP screening examination because of her premature birth at GA 29 weeks.

Results: Six children (4 males, 2 females) were included in the data analysis. First ophthalmic examination was performed between 3 days and 1.4 years of age. Normal visual development was present in all children. 4/6 children did not show any abnormalities of the anterior and posterior segment. Secondary glaucoma (glaucoma with non acquired systemic disease or syndrome) was diagnosed at age 4 months in one child who has an additional facial naevus anemicus. Subsequent trabeculotomy and deep sclerectomy treated her glaucoma successfully (last follow up age 4.4 years). Unusual and progressive retinal vascular abnormalities initially diagnosed as ROP like lesion were found in the one premature child. Anti VEGF injection was performed in the left eye because of progression. Initial regression was followed by repeated retinal hemorrhages in both eyes at age 2 weeks corrected age. Fluorescein angiography revealed an abnormal vasculature within the pars plana. Retinal hemorrhages resolved in both eyes.

Discussion: Severe and vision threatening ocular complications can occur in children with M-CM. Unusual vascular complications can occur, which are not reported at such an early age yet.

Conclusion: Early ophthalmic examinations are necessary to allow effective treatment if indicated. Differential IS IT COMPLETED?

Age-related normative values for visual fixation measured by a digital device

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Introduction: The ability to maintain gaze in an object is known as visual fixation (VF) and its development is progressive, depending on the definition of foveal structure and maturation of visual brain pathway. Good VF has to be accompanied with accurate saccades, which are fast movements to release the fixation from a point and direct it to another. Neurological conditions and ocular disorders have poor VF and defects in visual saccades. Hence, the examination of these abilities reports valuable information about the development of the visual system.

Remote eye tracking is a non-invasive technology, which requires very limited cooperation from children and have shown accurate results to measure VF. We present a new device (DIVE) for this purpose and its normative values.

Methods: Children younger than 14 years with no relevant medical records (except from mild ametropia) were examined by DIVE, which consists on a tablet with specific software to measure VF and an eye-tracker for gaze register. After a complete visual examination, participants underwent VF protocol at 50 cm of distance. The application began with a central stimulus, a high contrast target, followed by another peripheral one. The eye-tracker registered VF and saccades. For assessing fixation stability, the bivariate contour ellipse area was calculated, which quantifies the area of the ellipse containing most of the fixation positions. Fixational saccades were measured as saccadic reaction times (SRT) when foveal vision was shifted from the pursuit task to another point.

Results: The sample consist in 98 children. There were 5 groups according to age. Of them, 86 obtained valid SRT and in 92 the BCEA was calculated. The medium age was 5.9 years. VF improves with age but with not a significant increase (from 0 to 2 years 92 degrees squares in youngest children and 12 degrees squares in oldest, p=0.126), SRT improves with age with a significant increase (0.35 s in youngest children and 0.13 in oldest, p= 0.011).

Conclusion: Fixation and saccadic are normally subjectively assessed in children by clinical observation but currently technology may provide more accurate and objective assessment of oculomotor skills. We present age related normative values with our DIVE, which has demonstrated good reliability. Our device is portable and of easy use, so it allows accurate results and examinations with very few collaboration from the patient.
Retinal detachment in childhood is an uncommon but serious cause of visual loss. This lecture will focus on Rhegmatogenous retinal detachment (RRD) as the major cause of paediatric retinal detachment, with the majority of such cases being associated with inherited vitreoretinopathies.

Developmental abnormalities and trauma contribute most of the remainder and these will also be covered.

Clinical assessment with combined molecular genetic analysis is now routinely available to assist in the diagnosis and sub-classification of the inherited vitreo-retinopathies which in turn has facilitated risk assessment and strategies to reduce the incidence of retinal detachment in the high risk groups. This is especially important for children as once detachment has occurred the prognosis is often poor due to aggressive proliferative vitreoretinopathy (PVR) and late presentation with or without second eye involvement at the time of diagnosis.
Retinal haemorrhages in children

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Jaeger first described retinal hemorrhages in the newborn in 1861. This has been considered a benign, isolated finding with rapid spontaneous resolution the usual course. Following the Second World War, Caffey, an American pediatric radiologist, and Guthkelch, the first pediatric neurosurgeon in the U.K., described the “shaken baby syndrome” or “infant whiplash syndrome” as a specific form of child abuse in which retinal hemorrhages are a common finding. Careful examination of recent neuropathology findings in both these syndromes suggest that the original theses presented by these pioneering physicians require some revision.
Retinoblastoma and its management

Manoj Parulekar

Oxford Eye Hospital and Birmingham Children's Hospital - UK

Overview
Retinoblastoma is a cancer arising from the developing retina. This talk will address the genetics, early diagnosis, conventional and new management techniques of this highly treatable condition.

Genetics
Retinoblastoma may be heritable or non-heritable, and genetic testing is important. Screening of family members is essential for early detection. Lifelong surveillance of mutation carriers is needed due to the risk of second cancers.

Diagnostic modalities
The tumour is confined to the eye in the early stages, and cure rates for intraocular retinoblastoma can be as high as 95%. Extraocular spread carries a very poor prognosis with cure rates below 5-10%. Early diagnosis and prompt treatment is therefore crucial to save life and vision.

Management
Treatment requires significant multidisciplinary input, with local ophthalmic treatment, chemotherapy (systemic, intra-arterial or intra-vitreal) plaque radiotherapy, or surgery to remove the affected eye.

Newer treatment modalities including intra-arterial and intra-vitreal chemotherapy have significantly expanded treatment options, and many new treatments are currently being developed.
Other intraocular tumours in childhood

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The commonest paediatric intraocular malignancy is retinoblastoma but a wide range of intraocular tumours occur in childhood. Other retinal tumours include retinal capillary haemangioma and astrocytic hamartoma. The retina and retinal pigment epithelium can be affected by the combined hamartoma of these structures. The choroid can harbour a variety of tumours that includes haemangioma, both the circumscribed or diffuse types, osteoma or very rarely melanoma can occur in children. In the ciliary body, medulloepithelioma can be teratoid or non-teratoid, and can be benign or malignant. The presentation and management of these tumours is discussed.
First 22-months results of retinoblastoma management in a national reference center

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Introduction: Retinoblastoma is the most common ocular malignancy in pediatric population. In Portugal, the estimated incidence is approximately 8 cases a year. With the advent of new treatment modalities emerged the possibility of preserving more eyes and vision that would be otherwise compromised. In October 2015 the CHUC Ophthalmology department was recognized by the Ministry of Health as the sole national reference center in the treatment of ocular tumours. We aimed to report our centers’ experience in the management of retinoblastoma in a 22-months prospective analysis.

Methods: Prospective analysis of consecutive cases of retinoblastoma diagnosed and treated between July 2015 and April 2017 in our center. All included patients underwent a complete ophthalmological evaluation and fundus examination under general anesthesia. Demographic and clinical relevant data was recorded, namely age, gender and familiar history. Tumor dimensions, proximity to the optic nerve and macula, associated vitreous seeds were also documented and RetCam images were captured. The disease was staged according to the International Classification of Retinoblastoma. Depending on the disease stage, patients were treated with one or more of the following modalities: supraselective ophthalmic artery chemotherapy, intravitreal chemotherapy, endovenous chemotherapy, laser, cryotherapy and/or enucleation. Any adverse events possibly related to the treatment were also recorded.

Results: Twenty eyes of 16 patients were diagnosed with retinoblastoma. Predominantly male (62.5%), 4 patients had bilateral disease and 1 patient had pineal involvement. A mutation in Rb1 gene was found in 5 of the patients. Mean age at diagnosis was 22 months for unilateral cases and 17 months for bilateral cases. Eighty-one percent of cases presented with stage D, E and extracocular retinoblastoma. Nine eyes were enucleated due to advanced disease, 7 patients underwent systemic chemotherapy, 7 patients were treated with supraselective ophthalmic artery chemotherapy, 1 patient underwent intravitreal chemotherapy and 5 patients had adjunctive treatment with laser. The overall survival was 100% and no major events related to the treatment were registered.

Conclusion: Despite a good survival rate, ocular and sight survival results are still far from our expectations. With optimal treatment modalities already available, we defeat now the need to find new strategies to ensure an earlier diagnosis of the disease.

Outcomes following enucleation for Retinoblastoma

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Introduction: We report the long-term outcomes of children undergoing enucleation for retinoblastoma and results of porous polyethylene orbital implants.

Methods: Retrospective cases series. Included were all patients undergoing enucleation for retinoblastoma at BCH between 2002-2013. Patient demographics, implant characteristics, intra-operative details and postoperative complications were recorded.

Results: 239 children (127 female, 112 male) underwent enucleation for retinoblastoma over 11 years. Mean age at surgery was 2.3 years (range 0.2 - 12 years). Porous polyethylene (Medpor®) sphere implants were used in the majority of cases (93.6%) and the remainder had magnetic implants. The most common implant sizes were 20 mm (33.3%) and 18mm (30.7%). No significant intraoperative complications were encountered. The mean duration of follow-up was 36.5 months (range 2 weeks - 13.5 years).

8 patients (3.3%) developed orbital implant exposure, and were successfully repaired with dermis fat graft overlay or buccal mucous membrane A further 4 (1.6%) underwent implant exchange or replacement with fat grafts. 5 patients (2.1%) developed conjunctival prolapse and two patients (0.1%) developed granulation tissue in the socket. 5 patients (2%) required ptosis correction, and 4 (1.6%) underwent fornix reconstruction. Although some volume deficiency was common, most were successfully managed by prosthetists, and only 5 patients (2%) underwent volume augmentation with fat transfer or grafts.

Conclusion: Orbital implantation with porous polyethylene implants is associated with favourable outcomes following enucleation for retinoblastoma. Additional surgery was necessary in 10% of cases in this series. This number might increase with longer follow-up.
Rare case of permanent, bilateral severe visual loss secondary to Craniosynostosis in Alagille Syndrome

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**Introduction:** Alagille syndrome is an autosomal dominant, rare multisystem condition. Ocular involvement is generally non sight threatening. Craniosynostosis is a non-common association, which in a handful of cases has given rise to raised intracranial pressure and papilloedema, without significant visual loss. We report a unique case of bilateral, irreversible optic atrophy leading to permanent visual loss, secondary to Craniosynostosis in Alagille syndrome in a 5 year old child.

**Methods:** A four year old girl with Alagille Syndrome was referred by Paediatricians to Paeds Ophthalmology Clinic with concerns about deteriorating vision. The child had been born premature and had had ROP screening aged 36 weeks and 6 days and had been discharged, with ophthalmology review confirmed as normal in both eyes. The child had known cardiac and hepatic complications of Alagille syndrome, developmental delay and history of neglect, leading to subsequent adoption. She was otherwise a well and happy child.

**Results:** On exam, visual acuity was 3/24 with Cardiff Cards in both eyes. Nystagmus was noted on orthoptic exam and slit lamp exam revealed normal anterior segments but bilateral pale discs. An MRI head and visual pathways was requested to investigate causes of possible optic atrophy, which showed no mass, no haemorrhage or infarction but revealed cerebellar tonsillar ectopia, extending to 4mm below the level of the foramen magnum. Subsequent neurosurgical evaluation resulted in the child having a CT head, which showed pansynostosis and ICPM monitoring was performed, showing very high pressures up to 50 and A waves. The decision was taken to proceed with surgery and the child had successful calvarial expansion surgery a week after first neurosurgical review.

Follow up Ophthalmology review with Electrodiagnostic testing confirmed marked visual pathway dysfunction and the child was sight registered.

**Conclusion:** This case, to our knowledge, is the only case in the literature of such devastating visual loss in a child with Alagille Syndrome and highlights that the ocular complications in Alagille Syndrome can be serious. Alagille Syndrome is a multisystem condition and this child had regular cardiology and GI reviews. Although this child had a normal ocular exam at birth and there was history of neglect in her early years with late presentation to Ophthalmology, there is perhaps indication in Alagille Syndrome for more frequent ophthalmic exam to prevent visual loss.
Congenital craniofacial disorders are a diverse group of rare conditions which can pose complex management challenges. In syndromic craniosynostosis early fusion of the sutures of the calvarium, skull base and face causes midfacial hypoplasia with orbital distortion. This may result in exophthalmos, lagophthalmos, strabismus, amblyopia and ultimately blindness. Cranial distortion is associated with hydrocephalus and intracranial hypertension, which can present with papilloedema and lead to visual impairment. One of the key goals of the management of such patients is the preservation of visual function. In the more common non-syndromic craniofacial conditions severe ophthalmic abnormalities are less frequent, but expert ophthalmic evaluation plays a decisive role in identifying raised intracranial pressure when it occurs. Non-craniosynostotic disorders such as craniofacial clefting and Treacher Collins Syndrome very often involve the orbit and present specific reconstructive challenges. As such the ophthalmologist is a core member of the craniofacial multidisciplinary team.
Tales from the Children’s Toolbox

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Children are not small adults, and have a different spectrum of tissue activity and reactivity to adults, and these fundamental differences are evidenced in some of the unusual presentations of childhood orbital disease.

Through a collection of cases, the speaker will illustrate some unusual presentations of orbital disease in children and highlight the importance of certain clinical signs and symptoms.
Infantile capillary haemangioma (ICH) is the most common benign tumour in infancy, affecting about 1-4% of all infants, girls 3-5 times more often than boys and 23% of preterm babies of less than 1,000g birth weight.

Until 2008, local or systemic steroids were the main treatment form for ICH, but frequently at the price of significant side-effects. Following the serendipitous discovery of the remarkable therapeutic effects of beta-blockers, systemic propanolol has become the first-line treatment in severe or complicated ICH.

Since then, clinicians have begun to use topical beta blockers, in particular timolol maleate 0.5% solution, with good therapeutic effect, but fewer adverse reactions. Topical beta blockers are now used for lesions with both deep and superficial components and those that are amblyogenic. When initiated in the proliferative phase of the lesion, the effect of treatment may be seen within two weeks.

Several illustrative cases of deep periocular and orbital involvement will be presented as well as our first immunohistochemical and electron microscopical findings in ICH lesions after topical timolol maleate 0.5% treatment.
The retinal implant: from idea to reality

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Background: There exists no cure yet for blindness caused by hereditary retinal degeneration of the photoreceptors (e.g. retinitis pigmentosa; RP) but restoration of vision by various electronic retinal implants has made rapid progress in recent years with remarkable results concerning visual localization of objects, mobility, even reading and face recognition in some cases (Zrenner et al 2011, Zrenner, 2013). There are, in principle, three different approaches available to patients a) Epiretinal implants are positioned on top of the neuroretina, placing electrodes, controlled by a camera outside the body, at the functional output of the retina, i.e. near the RGC fibers (e.g. Humayun et al., 2012); b) subretinal implants that utilize light sensitive photodiodes, each connected to an electrode, both placed beneath the retina, contacting the functional input of the retina on the photoreceptor side (Stingl et al. Vis. Res. 2015) and c) the suprachoroidal approach that inserts electrode arrays from the back of the eye, positioning them on top of the choroid (Fujikado et al., 2011; Ayton et al., PLOS ONE, 2014).

Recent study (Stingl et al., Frontiers Neuroscience, Aug 2017): Safety and efficacy of a technically advanced subretinal electronic Retina Implant Alpha AMS device (Retina Implant AG, Reutlingen, Germany) was assessed in a prospective clinical multicenter trial in 15 patients with end stage retinitis pigmentosa (age 55.2 +/- 10.2 y, mean +/- SD). Functional outcome measures included 1) screen-based standardized 2- or 4-alternative forced-choice tests of light perception, light localization, grating acuity and Landolt C-rings; 2) grey level discrimination; 3) performance during activities of daily living (ADL-table tasks).

Results: Implant-mediated light perception was observed in 14/15 patients. During the observation period (12 months) implant mediated localization of visual targets was possible in 13/15 patients. Grating acuity was 0.1 cpd (cycles per degree) in 4/15; 0.33 cpd in 5/15; 1.0 cpd in 2/15 and 3.3 cpd in 1/15 patients. Best visual acuity assessed with Landolt C- ring was 20/546 and 20/1111. Of 6 possible grey levels, pairwise presented 3 times, 12 +/- 2.5 out of 18 were discerned correctly (mean +/- SD, n=10). Improvements (power ON vs. OFF) of ADL table tasks were reported by 13/15 patients. Results were overall stable during observation period. Serious adverse events (SAEs) were reported in 4 patients: 2 movement of the implant, readjusted in a second surgery; 3 conjunctiva erosion/dehiscence, successfully treated; 1 pain event around the coil, successfully treated; 1 partial reduction of silicone oil tamponade leading to distorted vision (silicon oil successfully refilled). The majority of adverse events (AEs) were transient and mostly of mild to moderate intensity.

Conclusions: Psychophysical and subjective data show that RETINA IMPLANT Alpha AMS is reliable, well tolerated and partially restores visual functions in the majority of patients. Surgical procedure is safe. Compared with previous Implant Alpha IMS, longevity of the new Implant Alpha AMS has considerably improved with a similar efficacy profile as Alpha IMS. Alpha AMS has meanwhile been certified as a commercially available medical device, in Germany reimbursed by the public health system. Providing centers have been recruited in several European countries.

Many more electronic devices are under development than could reasonably be presented here. A continuously updated list can be found at http://www.eye-tuebingen.de/zrenner/retimplantlist/

Literature: Consensus statements referred to here and references quoted can be found in a recent ARVO publication stemming from a Lasker Foundation meeting: at http://tvst.arvojournals.org/article.aspx?articleid=2212974
Congenital cataract and ectopia lentis: aetiology and new diagnostic pathways

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Ectopia Lentis and congenital cataract (CC) are uncommon but sight threatening conditions. Both conditions are largely caused by genetic mutations with some due to inborn errors of metabolism. Appropriate early intervention in cases amenable to treatment can not only maximize visual function but prevent systemic disease progression. Traditional diagnosis is a lengthy and costly process to health services and is typically unsuccessful - particularly in congenital cataract. Next generation sequencing (NGS) techniques are revolutionizing the utility of genomics and aiding precise diagnosis and subsequent provision of prognosis, management and treatment.

This talk will present the results of recent studies in this field and suggest models of care for children affected by these disorders.
Cataract surgery in children

Göran Darius Hildebrand

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Surgery for cataracts in childhood, especially in infancy, is much more complex than in adults and should therefore only be undertaken by experienced intraocular surgeons. We will look at the various standard techniques for opening the anterior and posterior capsules, removing the cataract and the multiple choices for aphakic and pseudophakic rehabilitation, starting at the time of surgery. The best outcomes are achieved with optimized timing and preparation of surgery preoperatively, meticulous surgical technique intraoperatively and careful management of amblyopia and any complications postoperatively.
Ectopia lentis and its surgical management

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Management of ectopic lens depends on many factors including the age of the patient, degree of lens displacement, status of the zonular apparatus, elasticity of the lens hardness, and some others. One of the key questions is whether or not to implant the IOL and what type of lens fixation method to choose. The decision is usually done on the strictly individual basis. When the patient’s age is exceeding 6 years our strong preference would be to preserve the capsular bag, and secure it to the scleral wall with the help of the modified capsular tension ring, and implant IOL in the bag. One of the critical steps of that procedure is anterior capsulotomies, which is very challenging due to the lens instability and lack of zonular tension to the capsule both making extremely difficult to initiate and complete the circular opening. In selected cases femtosecond laser can be used for anterior capsulotomy. Our preference is the mobile Z8 platform by Ziemer that is very useful in pediatric cases when the surgery is done under general anesthesia and patient traveling to the separate laser room is technically challenging. After capsulorhexis being completed, the specially designed capsule retractors help to stabilize the lens and facilitate the complete removal of it’s content. When securing the lens to the scleral wall the surgeon have to choose the appropriate suture material. Current choices are the 9-0 polypropylene or GoreTex (the latter is off-label). With 9-0 polypropylene our preference would be to utilize the Zig-zag suture technique for passing the needle 4-5 times parallel to the limbus and finally making a single locking suture. We used this technique for the last 8 years in over than 70 pediatric and adult cases of lens ectopia with great success and acceptable number of intra- and postoperative complications the most frequent being bleeding from the needle passing the ciliary sulcus and posterior capsule opacification.
The “Bag-in-the-lens” implant in pediatric cataract surgery

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The most frequent complications associated with pediatric cataract surgery are posterior capsular opacification (PCO) and visual axis reopacification (VAR). If the posterior capsule is intact the rate of PCO is up to 80%. A posterior capsulorhexis is therefore a standard step in pediatric cataract surgery. Despite this, lens epithelial cells (LECs) may still proliferate on the anterior hyaloid face resulting in VAR in up to 40% of pediatric patients. The “Bag-in-the-lens” (BIL) implant is a novel lens design that addresses the high risk of VAR in children.

The BIL is suspended by the anterior and posterior capsulorhexes and consists of a 5mm optic with two elliptical haptics, one anterior and one posterior, aligned 90° apart. When correctly sited, the lens itself acts as a mechanical barrier, restricting the access of proliferating lens epithelial cells to the posterior hyaloid preventing VAR. In our hands, the BIL maintains a clear visual axis and prevents VAR in 93.8% of pediatric patients. Failure to prevent VAR was due to peroperative inadequate BIL positioning. Thanks to peroperative OCT evaluation, this complication also can be avoided nowadays.
Visual outcomes five years following congenital and infantile cataract surgery with and without primary IOLs: findings from the IOunder2 study

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Introduction: Primary IOL implantation for congenital / infantile cataract has been widely adopted, despite the increased risk of secondary procedures, and uncertainties regarding visual benefit. Undertaken through the BCCIG, the IOunder2 study has established a (bi)nationally representative cohort of children under 2 years undergoing cataract surgery with or without IOLs, to investigate outcomes.

Methods: Children undergoing surgery identified and recruited through BCCIG; Prospective systematic data collection; Descriptive and multivariable multilevel regression analysis of visual outcome and associated factors.

Results: Findings based on 206/256 children for whom complete outcome data currently available IOL implantation in 57/130 children with bilateral cataract (BCC) and 37/76 unilateral (UCC). Overall, median age at surgery 7 weeks (IQR 5weeks-7months). Significant ocular co-morbidity in 42% BCC eyes ; 39% UCC eyes.

Visual outcome at 5yrs post-op (BCVA operated eyes): BCC: 0.5 logMAR (IQR 0.2-0.9, median BCVA BEO 0.38 logMAR)- UCC: 0.8 logMAR (IQR 0.4-1.5)

Visual outcome amongst children with ‘isolated cataract’ (without significant ocular co-morbidity) who underwent surgery in first 6 months of life:

BCC: 0.38 logMAR (0.25-0.625) IOL and 0.58 aphake (0.35-1.2)

UCC: 1.2 (0.375-1.4) IOL and 0.85 (0.6-1.2) aphake

Conclusion: IOLs do not appear to have conferred better visual outcomes on children with unilateral cataract, but may have resulted in improved visual outcomes following bilateral cataract surgery. The expected final size of the IOunder2 cohort follow up dataset will allow robust analysis of predictors of long term visual outcomes following surgery with or without primary IOLs.

Safety and outcome of paediatric cataract surgery

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Introduction: Paediatric cataract surgery is a complex procedure with multiple surgical risks and potential post-operative complications. Visual axis obscuration can be a particular problem, requiring simultaneous posterior capsulotomy and anterior vitrectomy at the time of surgery. Various lens positions have been advocated to maximise visual potential and reduce the need for re-operation. Furthermore, the rate of glaucoma post-operatively is estimated as high as 30%. We report our paediatric cataract surgery outcomes with particular reference to visual axis obscuration and glaucoma rate.

Methods: 48 cataract procedures were performed by the same surgeon (GDH) at two hospitals in the UK (John Radcliffe Hospital, Oxford and Royal Berkshire Hospital, Reading) between January 2010 and January 2016. A retrospective analysis of aetiology, change in visual acuity (VA), re-operation frequency and complication rate was performed.

Results: The average age at time of cataract surgery was 60.3 months (range: 1-190 months). Aetiology included unilateral and bilateral congenital cataract, trauma, and PFV-associated.

Significant peri-operative complications. An intraocular lens (IOL) was inserted at the time of lens aspiration in 75% of cases (36/48). 50% (18/36) of those undergoing primary IOL implantation had optic capture through both anterior and posterior capsulotomies. The mean VA in optic capture cases improved from 1.38 logMAR to 0.40 logMAR at a mean follow-up of 23 months (range: 1-55 months). Only 1/18 (5.6%) optic capture cases required further surgery, which was to clear proliferating Elschnig pearls. No case of posterior capsular opacification or anterior vitreous plaque formation was seen.

The incidence of post-operative glaucoma in the entire cohort was 8.3% (4/48): one case required goniotomy and one required goniosynechiolysis, both obtaining excellent IOP control without the need for long-term glaucoma medications. None of the four glaucoma cases were severe enough to require trabeculectomy or a tube procedure. There were no cases of endophthalmitis, retinal detachment, IOL decentration, hypopyon or persistent inflammation.

Conclusion: We demonstrate the general operative and post-operative safety and efficacy of the optic capture technique. Secondary glaucoma in this study was infrequent and did not require any filtering or tube surgery. The optic capture technique was associated with a very low re-operation rate of 5.6%.
**Bag-in-the-lens intraocular lens - well suited for use in children**

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**Introduction:** Visual axis opacification (VAO) is common after cataract surgery in children. The Morcher bag-in-the-lens (BIL) intra-ocular-lens (IOL) was created to prevent VAO. The aim of this abstract is to investigate complication rates in surgery for cataract with primary implantation of a BIL-IOL in children.

**Methods:** Medical records of 109 eyes of 84 children who had cataract surgery with primary BIL-IOL 2009 - 2013 were retrospectively reviewed for systemic or ocular comorbidity, postoperative complications and visual outcome. 42 were male (50%), 40 unilateral and 44 bilateral cataracts. Median age at surgery was 2.5 years (range 2 weeks - 14.1 years), 29 eyes (26.6%) of children were ≤3 months of age and 20 eyes (18.3%) 2 to 4 weeks of age. Median follow-up time was 2.8 years (7 months - 5.8 years). Exclusion criteria were uveitis or ≤6 months of follow-up.

**Results:** VAO was seen in 5/109 eyes (4.6%). Glaucoma in 15/109 (13.8%). One lens was exchanged due to myopic shift, one after traumatic dislocation, both easy to handle.

**Conclusion:** BIL-IOL is a safe lens in children. It prevents VAO formation even in the very young and is easy to exchange if the growth of the eye differs from what is expected.

**Glaucoma after cataract surgery in childhood - a report on 8 years of register based follow-up**

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**Introduction:** Glaucoma after paediatric cataract surgery is a well-known risk and a potential threat to vision. The aim of this paper is to present data on the occurrence of secondary glaucoma in children operated for cataract during eight years (2007-2014), in Sweden and Denmark, based on data from the PECARE, a subdivision of the Swedish National Cataract Register.

**Methods:** Data was derived from the Paediatric Cataract Register (PECARE), a bi-national web-based surgical register. Children with a cataract extraction who had surgery before 8 years of age between 2007 and 2014 were included.

On 31 December 2014, a total of 678 operated eyes were registered in 253 boys (52%) and 237 girls (48%), altogether 490 children. Excluded were glaucoma pre-operative, uveitis- or traumatic- caused cataracts and eyes that had surgery before 2007. The material thus consisted of 637 eyes in 239 boys and 215 girls. 299 eyes had a registered follow up at one year of age, 322 at two years and 260 at five years.

**Results:** 309 eyes 48.5% had surgery before one year of age. Overall 105 eyes of 637 had a registered glaucoma (16.5%). Only 6 eyes of 328 over the age of one had a glaucoma registered. Short axial length increased the glaucoma ratio in all eyes and persisting vascular membrane (PFV) increased the ratio in unilateral cataracts.

**Conclusion:** Early surgery for cataract increases the risk of early secondary glaucoma. Children who had surgery after one year of age rarely developed glaucoma.
Lensectomy reduces Schlemm canal diameter - a mechanical hypothesis of post-lensectomy glaucoma

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Introduction: Glaucoma remains one of the most serious long-term adverse events after childhood cataract surgery, and the underlying mechanism remains unknown. Chemical or inflammatory mediators may diffuse from the vitreous into the anterior chamber and induce trabecular meshwork (TM) inflammation and fibrosis. Alternatively, lensectomy may induce TM collapse or other aqueous drainage pathway changes. To date, there is no clinical evidence for either chemical or mechanical hypothesis. The aim of this work was to explore whether lensectomy is associated with a reduction in Schlemm canal (SC) and trabecular meshwork (TM) dimensions and an increase in TM density.

Methods: We acquired anterior segment OCT (AS-OCT) images of the nasal iridocorneal angle (ICA) of 50 children age 4 to 16 years with healthy eyes (mean age 10.9 +/- 3.0 years) and 48 who had undergone lensectomy (mean age 11.4 +/- 3.6 years), at two levels of accommodation (2.5D, 15D). In healthy volunteers, we analysed data from one eye per participant (left eyes); in children who had undergone cataract surgery, we used data from all post-lensectomy eyes (n=72 eyes). We used SPSS24 (IBM, Armonk, USA) for the comparison of all parameters (2-tailed independent samples t-test); a p-value under 0.05 was considered significant.

Results: The horizontal diameter of Schlemm canal and its cross-sectional area (SC-CSA) are significantly smaller in post-lensectomy eyes than in healthy eyes, both with relaxed accommodation and with accommodative effort; the vertical diameter is smaller with accommodative effort only. TM length is increased post-lensectomy, but we did not observe a significant difference in TM height or density between healthy and post-lensectomy eyes. In eyes which had developed glaucoma post-lensectomy, the horizontal diameter of SC and TM height were smaller than in eyes that had not developed glaucoma. There was no significant difference between eyes with glaucoma that had undergone trabeculectomy, tube implants or ciliary body laser ablation and those with glaucoma that had not undergone glaucoma procedures.

Conclusion: Lensectomy is associated with a reduction in diameter and cross-sectional area of Schlemm canal, which may reflect a reduction in outflow facility and contribute to the development of aphakic/pseudophakic glaucoma.

Simultaneous bilateral cataract surgery with IOL implantation in pediatric patients

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Purpose: To evaluate the safety and efficiency of simultaneous surgery and intraocular lens (IOL) implantation in pediatric patients with bilateral cataract.

Methods: Retrospective study including twenty bilateral pediatric cataract patients (40 eyes) that underwent cataract extraction and IOL implantation in both eyes in a single session, from 2004 to 2016. Patient age at surgery ranged from 3 months to 15 years (median 55 months). All operations were performed by a single surgeon and general anesthesia was administered to all cases.

Results: Mean follow-up time was 50 months. There were no cases of endophthalmitis or anaesthesia related problems. Only 1 intraoperative complication occurred (dropped nucleus, 1 eye with Marfan’s syndrome) among 40 eyes. Late postoperative complications include 2 eyes (5%) that developed glaucoma, 1 case of retinal detachment and 1 eye with pupillary membrane/posterior synechiae. Visual acuity (VA) was assessed in 32 eyes. Final best corrected VA was >20/40 for 25 eyes (78.1%), >20/200 and ≤20/40 for 6 eyes (18.8%) and no light perception (NLP) for 1 eye (3.1%).

Conclusion: Low rates of intraoperative complications suggest that simultaneous cataract surgery and IOL implantation could be a safe and efficient option for the management of bilateral pediatric cataracts. It offers the advantage of a single anaesthesia and potentially aids amblyopia management.
Orthoptist-led aphakia clinics
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Introduction: We have previously shown that routine use of the iCare rebound tonometer in measuring IOP allows easier awake examination for babies, infants and young children, and reduces the mean number of EUAs that a child undergoes per year. Although this creates an increase in the frequency of awake examinations needed, this has a positive impact on patient care and safety as patients require fewer general anaesthetics. Clinical experience with the iCare tonometer has allowed us to develop orthoptist-led aphakia clinics at our unit for the post-operative management and follow-up of aphakic children.

Methods: Retrospective case series. All patients attending the Birmingham Children’s Hospital Eye Clinic following surgery for congenital/developmental cataract in the years 2008, 2011 and 2016 were included. The orthoptist led aphakia clinics were introduced in 2012 and have been continuously audited since. At each visit, visual acuity, IOP using iCare and fundus photos are taken. The findings are assessed by the treating ophthalmologist, and clinician review arranged in the event of adverse findings. The total number of clinic visits, both to the orthoptist-led and ophthalmologist-led clinics, was recorded.

Results: There has been a >60% reduction in the number of EUAs, but a concomitant increase in clinics visits by more than 50%. The orthoptic led aphakia clinics have been successful in timely review of long term cataract follow-ups, reducing the pressure on ophthalmology clinics

Conclusion: iCare tonometry has enabled orthoptist-based aphakia clinics allowing safe and time-efficient long term follow-up of aphakic children.

Dynamic changes of iridocorneal angle morphology during accommodation in healthy children
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Introduction: Little is known about the possible link between developmental changes in amplitude of accommodation (AA) and intraocular pressure (IOP). In children under the age of 11 years, there is a negative correlation between AA and IOP; in young adults, sustained or repeated accommodative effort transiently lowers IOP. The aim of this study was to explore the mechanism linking accommodative effort and reduction of IOP; our hypothesis is that contraction of the anterior part of the ciliary muscle (CM) with accommodative effort may increase the diameter of Schlemm canal (SC) and induce morphological changes in trabecular meshwork (TM) and iridocorneal angle (ICA).

Methods: We acquired anterior segment OCT (AS-OCT) images of the nasal iridocorneal angle (ICA) and CM of 50 children age 4 to 16 years with healthy eyes, at two levels of accommodation (2.5D, 15D). We acquired semi-automated and manual measurements. We analysed data from one eye per participant (left eyes). We used SPSS24 (IBM, Armonk, USA) for the comparison of all parameters at two levels of accommodation (2-tailed paired t-test).

Results: With accommodative effort, vertical SC diameter increased significantly. In eyes with an increase in anterior CM thickness by 5% or more, trabecular meshwork length and standard ICA parameters (angle opening distance, trabecular iris space and trabecular iris surface area) also increased significantly with accommodative effort.

Conclusion: In children, physiological accommodative effort increases the vertical diameter of Schlemm canal; this may contribute to IOP regulation. AS-OCT may be of insufficient resolution to detect changes in trabecular meshwork morphology other than TM length.
Scheimpflug lens densitometry: an objective measure of cataract severity in children

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Introduction: Objectively grading the impact of lens opacities on children’s vision can be difficult. Pentacam Scheimpflug lens densitometry is an objective measure used in the management of age-related cataract; it is unclear whether this test would be useful in childhood cataracts, which are characterized by their varied morphology. The aim of this study was to evaluate testability of Scheimpflug lens densitometry in children and the association between these measurements and an established clinical severity grading and best-corrected visual acuity (BCVA) in children.

Methods: We enrolled 63 children, mean age 10.6, SD 3.2 years, with (n=45) and without (n=18) primary or secondary cataract, into this prospective, cross-sectional observational study. All underwent Pentacam Scheimpflug lens densitometry, clinician-grading of cataract severity, and BCVA measurement on age-appropriate logMAR charts.

Results: Lens densitometry measurements were successful in 56 of 63 eyes (89%). Repeatability was excellent for Pentacam Density Zone lens densitometry in the central 2mm of the crystalline lens (PDZ2mm), but poor for the automated Pentacam Nuclear Staging (PNS) score (Cronbach’s alpha 0.925 and 0.507, respectively). Average PDZ2mm was greater in children with cataract than those with clear crystalline lenses: median (interquartile range IQR) clear lens 8.75 (8.58 to 9.63), congenital/infantile cataract 10.10 (9.00 to 12.80), secondary cataract 9.20 (8.40 to 10.20). There was a strong association between PNS score and clinical severity grading (p<0.001), and PDZ2mm and both clinical severity grading and BCVA (p<0.001), but not PNS score and BCVA (p=0.201).

Conclusion: Despite their varied morphology and frequently anterior or posterior location within the crystalline lens, lens opacities in children can be detected by Pentacam Scheimpflug imaging, and their impact on vision can be estimated by the device algorithms. Further refinement or manual definition of a region of interest is needed to eliminate artefacts. This technique may be a useful adjunct to clinical assessment when deciding about the need for surgical intervention and for monitoring lens opacities for progressive opacification.

Is Ciclosporine 0.1% useful in children with severe ocular surface inflammation?

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Moorfields Eye Hospital NHS Foundation Trust, London - UK

Introduction: Ocular surface inflammation is common in children; chronic inflammation often requires continuous or frequent treatment with topical steroids, which can induce adverse events such as raised intraocular pressure and cataract. In 2015 a new topical preparation of ciclosporin A 0.1% became available (Ikervis, Santen Inc). The aim of this work is to present our experience with Ikervis in children, with emphasis on steroid-sparing impact and adverse events.

Methods: A search of pharmacy prescription records yielded a list of 60 children and adolescents who were first prescribed Ikervis between 15/06/15 and 01/12/16 at Moorfields. We reviewed the electronic records of these children.

Results: Mean age when Ikervis was first prescribed was 10.55 years (standard deviation SD 3.06); 25 children were girls. The commonest indication was vernal keratoconjunctivitis (n=34), followed by blepharokeratoconjunctivitis (n=15); other indications included allergic conjunctivitis (n=7), interstitial keratitis (n=2), ligneous pseudomembranous conjunctivitis and alkaline injury (both n=1). Median dosage was two drops per day. Mean duration of treatment to date is 29 weeks (SD 18). Topical steroids could be discontinued in 20 children. Three families discontinued Ikervis because of adverse events (stinging on application, lid swelling, skin irritation), two because of improvement in symptoms, and one because of compliance difficulties.

Conclusion: Our principal finding is that Ikervis allowed reduction of topical steroids in at least a third of patients; there were no serious adverse events, and tolerability is generally good. Review of the more detailed paper-based medical records is ongoing, and will allow more detailed evaluation of efficacy and safety.
Papilloedema or not?

David Taylor

University of London, London - UK

Papilloedema: raised ICP
- Clinical Diagnosis
- History
- Clinical Exam
- Tests

Congenital Anomalies
- “Glia” anomalies
- Developmental anomalies
- MGDA

Tumours & Infiltrations
- Leukaemias
- Combined hamartomas
- Gliomas
- Hamartomas
- Sarcoidosis
- TB

Drusen and other pseudo-papilloedemas
- Drusen
- Juvenile appearances
- Tests
- Unexpected presentations
- 15 second diagnosis

Myelinated Nerve Fibres
- Easy and difficult
- Hypertension
- Optic Neuritis/Neuro-retinitis
Space-occupying lesions in children may differ in various respects from those in adults. First of all, the space itself may be different: in infants, bulging of the fontanelles can relieve intracranial pressure that might otherwise produce papilledema. Or the infant skull may simply expand according to the tumour size or the secondary elevation in pressure it produces. For tumours such as craniopharyngiomas or gliomas which are present in utero, the masses themselves can alter and “mould” brain development such that even large tumours may not cause significant pressure points or handicap.

Differences in the cognitive nature as well as neural plasticity of young children also cause space-occupying lesions to present differently than in adult cases. Children rarely present when visual loss has been subtle or limited to one eye only. Nystagmus that is acquired and would cause intolerable oscillopsia in adults may produce no symptoms in the infant or toddler. Amblyopic suppression in a child will prevent the diplopia that would arise from even a subtle strabismus in adults.

While OCT and other forms of sophisticated ophthalmic imaging capabilities are ever improving, such devices are often ineffective in children unable to sit or cooperate sufficiently, and who may be unable to fixate. Objective assessment of neuro-ophthalmic signs of brain tumours remains essential to understand and assess a potential need for subsequent neuroradiologic imaging. While advances in neuroradiologic techniques permit us to better image tumours at earlier stages, these examinations nonetheless often require sedation in children and are not as easily ordered or risk free as for adults. When should one order imaging? How urgent must it be when sedation must also be planned? Do the potential benefits outweigh the risks? These are some issues that shall be addressed.
Paediatric tumours and their ophthalmic management in the paediatric ophthalmology clinic at a tertiary paediatric oncology centre

Guy Mole, Rachel Edminson, Caroline Hopper, Caroline Hildebrand
John Radcliffe Hospital, Oxford University Hospitals, Oxford - UK
Brighton and Sussex Medical School Postgraduate Medicine, Brighton - UK

Introduction: At a regional referral centre, a significant part of the Paediatric Ophthalmology workload involves looking after children with tumours of the head due to the clinical effects of the primary pathology or the surgery, chemotherapy or radiotherapy to treat it. The purpose of this study was to define the spectrum of tumours seen in our paediatric ophthalmology clinic.

Methods: A retrospective case review of the eye notes, clinic letters, radiology and histology reports, and multi-disciplinary meeting outcomes for all children with intracranial, orbital and facial tumours who had been under Paediatric Ophthalmology at our centre between 2012 and April 2017 was performed.

Results: There were 152 children (96 boys[63%], 56 girls[37%]) who fulfilled the inclusion criteria. 15/152 had died and 18 had been referred to their local unit for ongoing follow up. The range of age at presentation was 0 to 17 years (mean age 5.8 years).

The most common tumour types were astrocytoma(n=30, 20%), glioma(n=25,16%, medulloblastoma(n=15,10%), ependymoma(n=11, 7%), rhabdomyosarcoma(n=8, 5%) and craniopharyngioma(n=8, 5%). Of the children who died the most common tumour type was medulloblastoma(n=5), followed by astrocytoma(n=4) and ependymoma(n=2). The most common tumour locations were brainstem(n=35, 23%), multiple(n=21, 14%), diencephalon(n=20, 13%), visual pathways(n=17, 11%) and cerebellar(n=12, 8%).

Papilloedema was documented in 41(27%) and a further 13(9%) had symptoms of raised intracranial pressure or neuroimaging showing hydrocephalus. 99 children, which accounts for 72% of the children who are alive, are being managed for an active ophthalmic problem with the remainder mainly monitored for papilloedema. The most common reason children were seen was reduced visual field(n=32) then reduced vision(n=29). After these two the primary reasons for follow up were squint(n=12), corneal exposure(n=11) due to facial nerve palsy and diplopia(n=10).

27(6%) of the children underwent eye surgery of which the most common was squint surgery(n=11) followed by biopsy(n=4), tarsorrhaphy(n=4), phacoemulsification or lens aspiration(n=3) and enucleation(n=2).

Conclusion: This study is useful to understand which tumours are likely to present to the paediatric ophthalmology clinic and what ophthalmic manifestations commonly need to be managed. It also helps to shed light on the clinical course of the ophthalmic problems that arise from different tumour types and their prognosis.

A novel diagnostic next generation sequencing panel for infantile nystagmus

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Introduction: Infantile nystagmus (IN) is a genetically heterogeneous disorder arising from variants of genes expressed within the developing retina and brain. IN presents a diagnostic challenge and patients often undergo numerous investigations. We aimed to develop and assess the utility of a next-generation sequencing (NGS) panel to enhance the diagnosis of IN.

Methods: We identified 336 genes associated with IN from the literature and OMIM. NimbleGen Human custom array was used to enrich the target genes and sequencing was performed using HiSeq2000. Using reference genome material (NA12878), we show the sensitivity (98.5%) and specificity (99.9%) of the panel. We obtained saliva, extracted DNA and sequenced children with IN (n=40). These included diagnostically challenging cases where previous investigations were inconclusive often due to poor cooperation in children.

Results: We were able to identify the genetic diagnosis in 32/40 cases (80%). TYR and FRMD7 mutations were the most common cause. We identified rare variants within FRMD7 and CACNA1A intronic regions previously missed on exome sequencing (n=4). In one patient, copy number variation analysis revealed a FRMD7 deletion. The clinical diagnosis was revised in 6 cases (15%) based on the genetic data. In two cases this altered the clinical and surgical management. Genetic information from the panel will lead to personalised diagnosis, management and enable accurate genetic counselling in IN.

Conclusion: This is the first study establishing the clinical utility of a diagnostic NGS panel for IN. We utilised saliva samples as starting material, thus making it non-invasive and acceptable in the paediatric population. We show that the panel has high assay sensitivity and specificity. The genetic information from the panel will lead to personalised diagnosis, management and enable accurate genetic counselling in IN.

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John Radcliffe Hospital, Oxford University Hospitals, Oxford - UK
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Conclusion: This study is useful to understand which tumours are likely to present to the paediatric ophthalmology clinic and what ophthalmic manifestations commonly need to be managed. It also helps to shed light on the clinical course of the ophthalmic problems that arise from different tumour types and their prognosis.
Diagnostic pathway of optic atrophy in the paediatric population

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Introduction: The assessment of optic atrophy leading to diagnosis is challenging in the paediatric population. Optic atrophy is a descriptive clinical sign encompassing a spectrum of aetiologies including optic neuritis, congenital and hereditary neuropathies. The paediatric population needs specific considerations given examination constraints, preverbal children, secondary history from guardians and compliance with investigations. It is therefore important to appreciate the current paediatric patient population in order to plan services.

Methods: A retrospective chart review was performed in all cases of paediatric optic atrophy seen at Moorfields Eye Hospital London UK between 2010 and 2015. An electronic search of the hospital database was used.

Results: The search of the database “Open Eyes” revealed 43,497 records. Patients aged 16 years or younger identified 231 patients with a diagnosis of optic atrophy. Paper records of 155 patients were reviewed. Mean age for 155 patients were 7.54 years. 70 patients (30.3%) had neuroimaging (64 has an MRI and 4 had a CT scan). 105 patients had OCT scans of which 16 were abnormal (15.23%). 56 VEP studies were performed on patients. 50 were abnormal with 6 normal studies. 49 patients did not have electrodagnostics studies.

Conclusion: A diagnostic pathway is recommended for patients with suspected optic atrophy. This includes a detailed history and examination, paediatrician review, orthoptic assessment, optician refraction, photographs, OCT, EDTs and Neuro-Imaging (MRI scan) for first presentation patients.

Further investigations such as bloods should be tailored to the individual.

Involvement with multidisciplinary team i.e family support, CVI registration, counselling service and genetics service is recommended.

Imaging of the retina and optic nerve using optical coherence tomography in adolescents with surgically treated hydrocephalus

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Purpose: To map the morphology of the retina and optic disc in adolescents with in infancy surgically treated hydrocephalus and to compare the results with those of healthy controls.

Methods: Twenty-six adolescents with hydrocephalus (10 female, 16 male) with a median age of 15.0 years (range 13.3-16.9) and 31 sex- and age-matched controls underwent a detailed ophthalmological investigation including Optical Coherence Tomography (OCT). Assessed OCT parameters were: Macular retinal volume and thickness, thickness of the peripapillary retinal nerve fibre layer (ppRNFL), area of the optic disc, cup and rim as well as volume of the cup and rim.

Results: The average macular retinal thickness was found to be thinner amongst those with hydrocephalus compared with controls (p=0.0021 right eye (RE); p=0.0023 left eye (LE)) and the total retinal volume was found to be smaller (p=0.0003 RE; p=0.0014 LE). The hydrocephalus group also had a thinner total ppRNFL (p=0.0002 RE, p=0.0004 LE). However, optic disc, cup and rim areas or volumes showed no significant difference between the groups. Correlations were found between visual acuity (VA) and total ppRNFL (r=0.6, p=0.007 RE; r=0.6, p=0.0115 LE), and VA and disc area (r=0.6, p=0.016 RE; r=0.5, p=0.038 LE).

Conclusion: This study shows that there are significant differences in some of the assessed OCT parameters, e.g. the ppRNFL, between adolescents who underwent surgically treated hydrocephalus in infancy and controls. Following more research and clinical experience, OCT may be a useful method for early detection and follow-up of increased intra cranial pressure in individuals with hydrocephalus.
The usefulness of visual electrophysiology in pendular nystagmus in infancy

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Necker Hospital for Sick Children, Paris, France

Introduction: Strictly pendular nystagmus in infancy, partially or completely overlapping with so-called spasmus nutans-type nystagmus, has long been considered a benign entity. It is now a well-recognised symptom of various neurological or retinal diseases. In this study we looked at the place and usefulness of visual electrophysiology in the etiological work-up of pendular nystagmus in infancy.

Methods: All cases of infants having presented to our institution nystagmus clinic with a strictly pendular nystagmus from 2010 to 2016 were included. Infants with a causal diagnosis known before the first nystagmus clinic appointment were excluded.

Results: Fifty infants (34 boys, median age at nystagmus onset: 6 months) were included. MRI showed a large chiasmal glioma in 11 cases (22%), and a leukoencephalopathy in 5 cases (10%), a significant malformation in 3 cases (6%). Visual electrophysiology was realised in 44 cases (88%). ISCEV global ERG allowed to diagnose 17 cases (34%) of retinal dysfunction: cone dysfunction syndromes (8 cases), and early-onset severe retinal dystrophies (9 cases, 2 of which also exhibited a leukoencephalopathy).

Conclusion: In 68% of cases, pendular nystagmus was associated with a retinal or neurological disorder. In 50% of these symptomatic cases, visual electrophysiology was the key exam for diagnosing a retinal disorder. Unless a chiasmal glioma is found on MRI, a global ERG should systematically be part of the work-up of a pendular nystagmus in an infant, including in the presence of leukoencephalopathy.

The neuro-ophtalmic complications of childhood medulloblastoma and its treatment

Martin Wasik, Rachel Edminson, Göran Darius Hildebrand
John Radcliffe Hospital, Oxford - UK

Introduction: Medulloblastoma is the most common malignant tumour affecting the central nervous system in children. Although headaches, nausea, vomiting and ataxia are the predominant presenting features, ocular symptoms and signs are also frequently encountered. With better treatment regimes, the survival of patients with medulloblastoma is increasingly prolonged. It has therefore become more important to understand the mid- to long-term effects of the tumour and its treatment. We present our findings of the neuro-ophtalmic complications of this patient cohort from a single tertiary referral centre.

Methods: Children with a diagnosis of medulloblastoma being treated at the John Radcliffe Hospital, Oxford, were identified from our prospectively maintained database in the period January 2010 to April 2017. Patients notes were retrospectively reviewed.

Results: 32 patients were identified, of which 25 patients had undergone a post-operative orthoptic and ophthalmic assessment at least 6 months after surgery. 16% had ocular symptoms at presentation, while papilloedema was found in 61%. At follow-up, papilloedema had resolved in all patients, although a third of patients developed optic atrophy. Average post-operative visual acuity was good measuring 0.09 logMAR for each eye (range -0.2 to 0.6). 44% of patients had 6th cranial nerve palsies, 8% had 4th nerve involvement and there were no 3rd nerve palsies. 30% of patients with 6th nerve palsies underwent squint surgery. 8% were diagnosed with a horizontal gaze palsy, 8% with one-and-a-half syndrome, and 4% with skew deviation. 1 patient developed Parinaud’s syndrome post-operatively which resolved.

Conclusion: Neuro-ophtalmic complications are frequently encountered in children with medulloblastoma, especially after undergoing surgery. Overall the visual outcomes are encouraging, with very few children losing vision. It is crucial that these patients receive ongoing ophthalmic follow-up in order to preserve vision, manage diplopia and ensure adequate ocular surface protection.
Neuro-ophthalmic features of paediatric diencephalic tumours

Zuzana Sipkova, Guy Mole, Göran Darius Hildebrand

Oxford Eye Hospital, John Radcliffe Hospital, Oxford - UK

Introduction: We report on our experience of neuro-ophthalmic and clinical characteristics of children with diencephalic central nervous system tumours.

Methods: A retrospective case review of paediatric patients with history of diencephalic tumour involving the thalamus, hypothalamus and pituitary gland who were monitored at the Oxford Eye Hospital between 2012 and 2017 was performed.

Results: Twenty-seven patients under the age of 16 years were reviewed, comprising 17 male and 10 female patients. The age range at presentation was 0-15 years (mean age 6.3 years). The most common tumour location was the pituitary gland (n=12, 44%) followed by the hypothalamus (n=5, 19%) and thalamus (n=3, 11%) and pineal gland (n=3, 11%). Four patients had tumours involving multiple locations, including the diencephalon.

Histological confirmation of diagnosis was available in 18 patients and included craniopharyngioma (n=7), pilocytic astrocytoma (n=5), germ cell tumour (n=2), neuro-glial tumour (n=2), ganglioglioma (n=1) and lymphoma (n=1).

Significant visual field defect was present in 11 patients with five having recorded bitemporal hemianopia. Reduced vision (best corrected visual acuity worse than LogMar 0.3 in either eye) was recorded in ten patients (37%) and three patients had no perception of light in one eye (severe optic atrophy in two cases and enucleation for retinoblastoma in one case). The most common systemic clinical presentation included symptoms of raised intracranial pressure (n=11, 41%), including headaches (n=8), personality change (n=2) and nausea and vomiting (n=2). Papilloedema was noted in seven cases and two had sixth cranial nerve palsy. Other ophthalmic signs included Parinaud’s syndrome (n=1), nystagmus (n=3) and optic pallor/atrophy (n=11). Neurofibromatosis type 1 was diagnosed in 3 cases (11%). Diencephalic syndrome, manifested by failure to thrive and progressive emaciation, was noted in one patient.

Conclusion: Significant number of children with diencephalic tumours presented with systemic symptoms of raised intracranial pressure. Signs of papilloedema and optic nerve pallor or atrophy were common on ophthalmic examination.

Ophthalmic complications of proton beam radiotherapy for paediatric patients with intracranial and orbital tumours

Kavita Aggarwal, Fabienne Fierz, Shaun Wilson, Kate Wheeler, Göran Darius Hildebrand

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Introduction: Radiation therapy has known ocular complications which is reported mainly in adult population. With the introduction of proton beam radiation therapy (PBRT), a conformal technique that decreases the dose to the normal brain tissues, the risk of late complications may be decreased. PBRT is becoming increasingly used especially in treating paediatric brain tumours although unfortunately there is limited data at present on ophthalmic complications (1,2). We report the incidence of ophthalmic complications in 18 paediatric patients treated with PBRT.

Methods: A retrospective review of medical records of all paediatric patients presenting to Oxford University Hospitals NHS Foundation Trust with primary orbital or intracranial malignancies who were treated with PBRT between 2011-2017 was carried out. Ophthalmological and paediatric data pre and post PBRT was extracted.

Results: 18 patients were identified. The median age at diagnosis was 7 years (range 0.5-13.5 years) with a median follow-up time after PBRT treatment of 44.5 months (16-102 months). 13 (72.2%) had intracranial tumours including craniopharyngioma, ependymoma and astrocytoma. 4 (22.2%) had orbital tumours and 1 (5.6%) had a mandibular tumour, all of which were rhabdomyosarcomas.

In all patients there was no significant difference in pre and post PBRT visual acuity. No clinical deterioration of pre-existing optic atrophy, visual field defect, eye movement deficits and nystagmus was recorded.

All four patients treated for orbital tumours had anterior segment complications including dry eye, corneal vascularization and were managed with long term lubrication and one patient developed ptoxis post treatment. Two patients required cataract surgery and further YAG capsulotomy. Patients following cataract surgery had excellent visual outcomes. None of the patients developed secondary glaucoma or radiation retinopathy during the follow-up period.

Conclusion: Ocular complications following PBRT for intracranial malignancies were rare in our cohort but longer follow up may identify these (1,2). However in orbital tumours there is a higher risk of ocular complications requiring treatment including surgery, although final visual outcome was good. As PBRT is becoming more readily available, larger and longer term data will need to be analysed.

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The Heimann-Bielschowsky phenomenon: a pediatric case series

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Introduction: The Heimann-Bielschowsky phenomenon (HBP) is a monocular, pendular, vertical nystagmus characterized by a low frequency and a low amplitude, in an eye with profound visual loss. It has been classically considered as an adult-only type of nystagmus. We describe a series of infants with HBP and characterize this ocular motor disorder by video- and/or infrared oculography.

Design: Retrospective observational case series.

Methods: Infants examined in the pediatric neuro-ophthalmology clinic between 2009 and 2017 and having presented with HBP were retrospectively collected.

Results: Six cases (1M : 5F) were included. Age at HBP presentation ranged from 3 to 112 months. All cases had visual acuity worse than 0,16 in the affected eye (4 right and 2 left eyes). Strabismus was seen in three cases (2 divergent and one convergent strabismus). No one had symptoms of diplopia or oscillopsia. All cases had unilateral, slow, pendular vertical oscillations occurring in the amblyopic eye. Video-oculography showed frequencies lower than 5Hz with amplitudes lower than 15°. Three infants had visual pathway gliomas with unilateral severe optic atrophy, one had optic nerve astrocytoma associated with tuberous sclerosis, one had post-traumatic third nerve palsy and optic nerve atrophy and the last had persistent hyperplastic primary vitreous complicated by complete retinal detachment.

Conclusion: HBP may be observed in children and infants. Its relations with spasms nutans-type of nystagmus, the main variety of pendular dissociated infantile-onset nystagmus, are not clear. It is nowadays probably still underdiagnosed and its physiopathology is not well understood. It could result from disruption of the fusional vergence or of the monocular visual stabilization systems.

Retinal imaging as an objective measure of ocular disease in Mucopolysaccharidosis

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Introduction: The mucopolysaccharidoses are a group of rare lysosomal storage disorders. Ocular features include corneal clouding, retinopathy, glaucoma and optic neuropathy. Visual impairment has a huge impact on quality of life. There is limited knowledge of the effect that systemic treatments have on the ocular features of this disease. Available evidence is largely based on small retrospective studies and subjective measures of eye disease. Retinal imaging may provide more objective measures of the severity and progression of retinopathy and optic neuropathy.

Methods: Prospective observational study. Patients recruited from paediatric ophthalmology clinics at Manchester Royal Eye Hospital. Heidelberg OCT (Spectralis) and wide-field Optos Vantage imaging were performed in all patients when possible.

Results: OCT macula was performed in 12 patients (10 MPSI, 1 MPSIV & 1 MPSVI). Mean central foveal thickness and central subfield thickness (EDTRS 1mm), were similar to previously published values for healthy individuals. 2 MPSI patients showed evidence of parafoveal photoreceptor loss, one of whom had no clinical suspicion of retinopathy. Another MPSI patient had widespread thinning of photoreceptor layers and a hyporeflective foveal lesion in his left eye. Some degree of foveal external limiting membrane (ELM) thickening was noted in 9 MPSI patients. 1 MPSI patient showed evidence of significant chorioretinal folds. Optic nerve OCT was done in 8 patients and were in-keeping with individual other clinical features.

Optos wide-field imaging was performed in 12 patients (9 MPSI, 1 MPSIV & 2 MPSVI). Imaging showed clinically detected optic nerve abnormalities and pigmented retinopathy, although image quality varied with the level of corneal clouding present. In 1 MPSI patient peripheral RPE mottling was noted that had not been reported clinically. Interestingly, 1 patient with known ERG evidence of rod cone dystrophy had unremarkable Optos imaging.

Conclusion: These findings suggest OCT and Optos imaging can be useful in detecting and monitoring retinopathy, cystoid macular oedema and optic nerve abnormalities in patients with MPS. Objective monitoring of ocular features over time will allow better understanding of the effects of systemic treatments.
Vision and visual perception in adolescents with hydrocephalus—a long-term follow-up

Susann Andersson, Johanna Norström, Marita Andersson Grönlund

Department of Clinical Neurosciences, Institute of Neuroscience, Sahlgrenska University Hospital, Gothenburg, Sweden

Introduction: The incidence of congenital hydrocephalus (HC) is estimated to approximately 0.7-1.1 children per 1000 live births. Symptoms vary; cerebral palsy, epilepsy and learning difficulties are common as well as visual deficits. Almost 60% have visual perceptual problems (VPP) which means difficulties with interpretation of visual input. The aim of the study was to investigate visual acuity (VA) and VPP over time in a group of individuals with HC surgically treated in infancy, and compare the results with a healthy control group. Furthermore, to investigate whether there is a correlation between VA and VPP or not.

Methods: Twenty-six adolescents (10 female, 16 male), born 1999-2002 (median age 15.0 years), with HC were studied. Visual acuity was tested and VPP were evaluated through structured history-taking. The results were compared with data from participants’ childhood (median age 8.7 years) and with an age- and sex-matched control group (n=31).

Results: Fifteen out of 23 (61%) adolescents reported VPP, 12/28 (43%) in childhood (n.s.) and 2/31 (6.5%) in the control group (p<0.0001). The median VA in childhood was 0.9 decimal (fix-1.25) and 1.0 decimal (fix-1.25) in adolescence compared with 1.25 (1.0-1.25) in healthy controls (p<0.0001). No correlation between VA and VPP was found.

Conclusion: Our results indicate that children with HC maintain the frequency of VPP in adolescence. Approximately 60% reported VPP, compared with 6.5% in healthy controls. A wide range in VA was noted - some adolescents with HC could only fixate while others had a normal VA. However, no correlation between VA and VPP was found.
Optic Nerve Sheath Fenestration (ONFS) in Paediatrics

John Elston
Oxford, UK

The rationale of this intervention is to decompress the optic nerve sheath “compartment” - the subarachnoid space surrounding the orbital and intra-canalicular optic nerve. It is most often used to stop or reverse visual loss secondary to chronic papilloedema in idiopathic intracranial hypertension, IIH. ONSF has also been advocated in traumatic optic neuropathy.

Three techniques are recognised: trans-conjunctival, trans-cutaneous and endoscopic ONSF.

The outcome of ONSF in adults has never been the subject of any form of clinical trial. The available data is class III evidence. Few paediatric series have been published.

Multiple retrospective reports indicate that it is a safe and largely effective intervention. Individual surgical expertise is important; total or near total blindness has been reported.
SATURDAY 2 SEPTEMBER

Neurosurgery for raised ICP

Shailendra Magdum
Oxford Children's Hospital - UK
POSTER LISTING
FRIDAY 1 SEPTEMBER

F1) B. Urban, A. Bakunowicz-Łazarczyk, D. Średzińska-Kita POLAND
Retinal changes associated with long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency

F2) C. Blanco-Angel, N. Palmou, S. Miranda SPAIN
Panuveitis in a 9-year-old boy with aggressive ulcerative colitis treated with adalimumab: a case report.

F3) C. Gurez, B. Gökyığit, M. Taşkapılı TURKEY
Macular and Retinal Nerve Fiber Layer Thickness in Amblyopia

F4) N. Fomina, E. Saidaşheva RUSSIA
Aniridia as a main sign of WAGR syndrome

F5) C. Cassiman, F. Vermeulen, C. Wouters, L. De Somer, J. Van Calster, I. Casteels BELGIUM
Bilateral idiopathic macular choroidocapillarisitis in an otherwise healthy 11 year old boy.

F6) K. Ozols, A. Treija, S. Valeina LATVIA
Accidental retinal finding can sometimes be lifesaving.

F7) M. Grudzinska Pechhacker SWEDEN
Novel mutations of the RDH12 and MERTK gene in a family with early onset retinal dystrophy

F8) S. Hussain, M. Iveson UK
Case Report: An Indicentlal Finding of Severe Familial Exudative Vitreoretinopathy

F9) A. Rigaudy UK
Situational Analysis of Paediatric Tertiary Ophthalmology facilities in African VISION 2020 LINKS

F10) A Aligera, S. Valeina LATVIA
Case Report of acute posterior multifocal placoid pigment epitelioathy

F11) H. Attila, F. Tüntäş-Bilen, H. Attila TURKEY
Optical Coherence Tomography Angiography Findings in Leber Congenital Amaurosis

F12) H. Attila, P. Bingöl Kuzlentung, H. Attila TURKEY
OCT Angiography Findings in Leber Hereditary Optic Neuropathy

F13) N. Karîmînâva UZBEKISTAN
Analysis of haemodynamically parameters of ocular and renal arteries in children with chronic glomerulonephritis

F14) K. Palumaa ESTONIA
X-linked retinoschisis – a case report of genetically proven X-linked retinoschisis of two brothers

F15) R. Parness Yossifon, B. Hadas, H. Leiba ISRAEL
Asymptomatic autosomal recessive bestrophinopathy: Can it be treated?

F16) A. Houtman THE NETHERLANDS
Toying with the epiCam: enhancing the view in infants

F17) R. Parness Yossifon, H. Leiba, B. Hadas ISRAEL
Foveal hypoplasia: Normal does not always mean normal and abnormal is not always abnormal

F18) T. Kuhn, H. Katus, J. Krijgs veld, F. Leuschner GERMANY
Novel secretome analysis to identify crucial signaling proteins in the cellular communication

F19) S. Valeina, V. Sperga, I. Orube LATVIA
Case report: Hypertensive retinopathy

F20) S. Oto, A. Sangüi Sezenôz, İ. Akkoyun, S. Bayar Akça, G. Yılmaz TURKEY
OCT findings and visual prognosis in children with unilateral myelinated retinal nerve fibers and high myopia

F21) S. Defoort-Dhellemmes, I. Drumare, V. Smirnov, B. Puech FRANCE
SD-OCT and FAF at onset of inherited maculopathy in children

F22) N. Voide, F. Munier SWITZERLAND
Torpedo maculopathy: a multimodal analysis of a pediatric case series

F23) V. Pueyo, J. Ignacio Echeverría, K. Plunkett SPAIN
Validity and usability of new stimuli for colour vision assessment in infancy

F24) A. Dahlmann-Noor, M. Crossland, H. Unwin, V. Gothwal, R. Thomas UK
Assistive Technology for children with low vision: the CREATE pilot trial (Children Reading with Electronic Assistance To Educate)

F25) R. Chadha UK
10 year audit of The Paediatric Low Vision Aid Clinic at Oxford Eye Hospital

F26) M. Ali, D. Tracey-White, M. Smart, A. Webster, M. Moosaje UK
Combined nonsense-mediated decay inhibition and translational readthrough as a treatment approach for hereditary retinal dystrophies

F27) E. Larsson, B. Carlsson, C. Lundberg, G. Holmström, H. Åkerblom SWEDEN
Different phenotype in three siblings with a mutation in the ABCA4 gene – a case report.

F28) A. Monnier, O. Michel, D. Zambrowski, D. Bremond Gignac, M. Robert FRANCE
Post infectious pigmentary retinopathies in infants

F29) N. Osipova, E. Denisova, L. Kogoleva, O. Novikova, N. Osipova RUSSIA
Choroidal neovascularization secondary to best vitelliform macular dystrophy: is it rare?

F30) M. Jarc-Vidmar, B. Štirm-Kranjc, P. Jaki-Mekjavič SLOVENIA
Intravitreal anti-VEGF treatment for choroidal neovascularisation secondary to best vitelliform macular dystrophy in a 5 year old child and his grandmother

F31) A. Majander, E. Sankila, K. Vasara, A. Haavisto, S. Seitsonen, K. Avela FINLAND
A TULP1 mutation in Finnish patients with congenital nystagmus and early onset retinal dystrophy (EORD)

F32) V. Smirnov, I. Drumare, S. Defoort-Dhellemmes FRANCE
Hereditary retinal diseases diagnosed in infants with congenital vertical nystagmus.

F33) G. Yılmaz TURKEY
Severe ocular complications in patients with macrocephaly-capillary malformation syndrome
M. Kowalczyk, R. Rejdak  POLAND
Autosomal dominant neovascular inflammatory vitreoretinopathy

E. Maka, A. Szigeti, M. Bausz, M. Resch, Z. Zsolt Nagy  HUNGARY
Is ROP the worst abnormality what we can find in a preterm baby?

A. Gerasimenko, V. Krasilnikova  BELARUS
The macula zone status in cicatrical period of retinopathy of prematurity according to optical coherence tomography

A. Gerasimenko, V. Krasilnikova  BELARUS
Late retinal detachment in adult retinopathy of prematurity

G. Reid, M. O’Neill, E. McLoone  UK
Retinal injures resulting from handheld laser devices in the paediatric population, A case series of 27 eyes.

T. Pérez Roche, E. Prieto, O. Castillo, J. Gutierrez, M. Ortin, D. Gutierrez  SPAIN
Age-related normative values for visual fixation measured by a digital device

J. Providência, G. Castela, S. Silva, E. Machado, J. Murta  PORTUGAL
First 22-months results of retinoblastoma management in a national reference center

P. Shah, F. Shah, M. McCalla, Z. Squires, M. Parulekar  UK
Outcomes following enucleation for Retinoblastoma

L. Derriman, M. Ayoubi, A. Channa, I. Gout, A. Toufeeq  UK
“Rare case of permanent, bilateral severe visual loss secondary to Craniosynostosis in Alagille Syndrome”

D. Giannoulis, E. Kostopolou, A. Chranioti, A. Mataftsi, N. Ziakas  GREECE
Simultaneous bilateral cataract surgery with IOL implantation in pediatric patients

P. Shah, L. Ramm, M. Awad, C. Dewsbury, M. Parulekar  UK
Orthoptist-led aphakia clinics

Dynamic changes of iridocorneal angle morphology during accommodation in healthy children

Scheimpflug lens densitometry: an objective measure of cataract severity in children

E. Papamichael, S. Parvizi, K. Gadthi, P. Vetpillai, M. Hingorani, A. Dahlmann-Noor  UK
Is Ciclosporine 0.1% useful in children with severe ocular surface inflammation?

R. Karim, D. Oladiwura, A. Dahlmann-Noor, A. Sawczenko  UK
Diagnostic pathway of optic atrophy in the paediatric population

M. Andersson Grönlund, L. Mybeck, S. Andersson  SWEDEN
Imaging of the Retina and Optic Nerve Using Optical Coherence Tomography in Adolescents with Surgically Treated Hydrocephalus

The usefulness of visual electrophysiology in pendular nystagmus in infancy

M. Wasik, R. Edminson, G. Darius Hildebrand  UK
The neuro-ophthalmic complications of childhood medulloblastoma and its treatment

Z. Sipkova, G. Mole, G. Darius Hildebrand  UK
Neuro-ophthalmic features of paediatric diencephalic tumours

K. Aggarwal, F. Fierz, S. Wilson, K. Wheeler, G. Darius Hildebrand  UK
Ophthalmic complications of proton beam radiotherapy for paediatric patients with intracranial and orbital tumours.

N. Voide, M.P. Robert  SWITZERLAND
The Heimann-Bielschowsky phenomenon: a pediatric case series
S32) R. Parness Yossifon, H. Leiba, B. Hadas *ISRAEL*
Long term ophthalmic follow-up in a case with long-chain 3-hydroxyacyl-coa dehydrogenase (LCAD) deficiency

S33) O. Pfäffli, C. Gerth-Kahlert *SWITZERLAND*
Color vision testing in young children: performance comparison of normal and impaired vision subjects

S34) S. Shidik, M. Sidik *INDONESIA*
Optic Neuropathy in Children: A case series

S35) S. Stevenson, S. Downes, C. Andrews, K. Wulff, I. Alexander, R. Foster *UK*
The impact of optic nerve disorders on sleep wake

S36) H. Akerblom, E. Larsson, G. Holmström *SWEDEN*
Pre-term birth affects the optic nerve morphology in school-aged children.

S37) Z. Sipkova, G. Darius Hildebrand, J. Norris *UK*
Acute sickle cell orbitopathy masquerading as orbital cellulitis

S38) P. Terry, V. Joganathan J. McMahon, S. Wall, J. Norris *UK*
Non-surgical management of craniofacial dystopia through specialist scleral contact lenses in combination with prismatic glasses

S39) Š. Markelj, G. Markelj, B. Štim Kranjac, M. Tekavčić Pompe *SLOVENIA*
Title: Rare cause of bilateral optic disc oedema in a 2-year old girl

S40) I. Knisbacher, A.L. Marcovich, C. Vinkler, M. Huszar, H. Leiba *ISRAEL*
Bilateral inferior corneal pseudopterygium in Kabuki syndrome

S41) J. Szczapa-Jagustyn, A. Gotz-Więckowska *POLAND*
Ophthalmic findings in pediatric patients with chronic autoimmune thyroiditis

S42) K. Aggarwal, F. Ryan, J. Jayamohan, M. Gurnell, G. Darius Hildebrand *UK*
Sustained effect of somatostatin analogue treatment on visual recovery in a 13-year-old boy with advanced thyrotrophic pituitary macroadenoma

S43) V. Pueyo, I. Gonzalez, V. Pueyo, E. Prieto, T. Perez, I. Altemir *SPAIN*
Visual perceptual disabilities in children adopted from Eastern Europe

S44) E. Prieto Calvo, I. Gonzalez Viejo, T. Pérez Roche, O. Castillo Castejón, A. Fanlo Zaragoza, B. Masia *SPAIN*
A quantitative study of fixation stability in amblyopia

S45) A. Bakunowicz-Łazarczyk, B. Urban *POLAND*
Complications of retained intraorbital wooden foreign body

S46) P. Delbeke, J. Willemot, I. Balikova *BELGIUM*
Corneal microcysts due to Cytarabine toxicity

S47) D. Makhkamova *UZBEKISTAN*
Causes of congenital optic nerve abnormalities

S48) R. Jones, J. Soo, A. Dahlmann-Noor, O. Jeelani, A. Sawczenko *UK*
Ophthalmologists remember Rickets: Nutritional Vitamin D Deficiency causing severe visual impairment in a child

S49) R. Karim *UK*
Non-surgical interventions for nystagmus developing in the first year of life (infantile nystagmus)

S50) R. Karim, S. Tuft, F. Larkin *UK*
Paediatric Vernal Keratoconjunctivitis impact of on quality of life using Quick Questionnaire

S51) S. Odent, S. Le Piane, C. Cassiman, I. Casteels *BELGIUM*
A bubble in the eye?
POSTER ABSTRACTS
Asymptomatic autosomal recessive bestrophinopathy: Can it be treated?
Noa Gilead, Beatrix Hadas, Hana Leiba, Reut Parness Yossifon
Kaplan medical center, Rehovot - Israel

Introduction: Autosomal recessive bestrophinopathy (ARB) is a disorder caused by biallelic mutations in the BEST1 gene. It has a variable phenotype with extramacular multifocal yellowish subretinal deposits, extensive subretinal fluid, and cystoid macular edema. Carbonic anhydrase inhibitors have been used in the treatment of cystoid macular edema in several other retinal dystrophies such as retinitis pigmentosa and juvenile retinoschisis.

Purpose: To describe the clinical findings in an 8 years old boy with autosomal recessive bestrophinopathy treated with topical carbonic anhydrase inhibitors (CAI).

Methods: We present the case of a patient with ARB including a complete ophthalmic examination, fundus photography, spectral-domain optical coherence tomography (SD-OCT), fluorescein angiography, fundus autofluorescence imaging, full-field electroretinography (ERG) and electro-oculography (EOG).

Results: An asymptomatic 8 years old boy with 6/6 visual acuity has been found to have multiple bilateral extramacular yellowish subretinal deposits. On further investigation, SD OCT demonstrated a shallow serous retinal detachment throughout the macula, hyper-reflective subretinal deposits, elongation of photoreceptor outer segments and cystoid macular edema. The subretinal deposits were hyperfluorescent on fundus autofluorescence imaging. Photopic and scotopic responses on full-field ERG were normal and on EOG the Arden ratio was reduced. Due to the presence of subretinal fluid and cystoid macular edema he has been treated with Dorzolamide three times a day. SD OCT was repeated after treatment with topical CAI.

Conclusion: We present a case of an asymptomatic 8 years old boy incidentally diagnosed with autosomal recessive bestrophinopathy. We discuss the natural history of ARB, the necessity of treatment in asymptomatic case and the result of treatment with Dorzolamide.
Panuveitis in a 8-year-old boy with aggressive ulcerative colitis treated with adalimumab: a case report

Carolina Blanco-Angel, Natalia Palmou, Silvia Miranda
Laredo Hospital, Laredo - Spain
University Hospital Marques de Valdecilla, Cantabria - Spain

Introduction: Data on ocular manifestations of inflammatory bowel disease (IBD) in children are limited. Some authors have reported successful results with adalimumab therapy in children with uveitis due to IBD. But there are no recommendations on which ophthalmologic treatment and follow-up should be offered to children with uveitis associated to ulcerative colitis (UC).

Methods: We describe an 8-year-old boy with total colectomy and ileostomy due to an aggressive UC and a history of recurrent panuveitis in the right eye. He developed with loss vision, eyelid swelling, acute anterior uveitis and cystoid macular edema (CME). Serological determination for HLA B27, FR and ANAs were negative.

Visual acuity, anterior chamber cell grade and CME measure by Optical Coherence Tomography (OCT) were assessed during treatment.

Results: The patient was treated with both oral and topical glucocorticoid. In order to minimize corticoid- associated side effects he was also treated with adalimumab (25mg/15 days) and corticosteroid dose was progressively reduced.

Clinical manifestations were recorded 1, 3, 6 and 9 months after treatment. Visual acuity improved from 20/70 to 20/20, anterior chamber inflammation disappeared and central foveal thickness decreased from 870 m to 280 m. After 9 months of treatment the patient remains asymptomatic.

Conclusion: There is a clear demand for additional effective and steroid-sparing therapies in paediatric population with non-infectious uveitis. Adalimumab is indicated for patients 6 years of age and older with other IBD as Crohn’s disease. It could be an effective agent in off-label treatment of paediatric uveitis associated to ulcerative colitis as well.

Macular and retinal nerve fiber layer thickness in amblyopia

Ceren Gürez, Birsen Gökylięt, Muhittin Taşkapılı
University of Health Sciences Beyoğlu Training and Research Hospital, Istanbul - Turkey

Introduction: This study was performed to measure the retinal nerve fiber layer (RNFL) and the macular thicknesses using optical coherence tomography (OCT) in children with unilateral amblyopia.

Methods: Measurement of the retinal nerve fiber layer and macular thickness for both amblyopic and normal fellow eyes carried out by OCT.

Results: Fifty six patients with unilateral amblyopia between the ages of 4 years and 10 years were included. The macular retinal thickness and the RNFL thickness were measured using OCT. The mean macular retinal thickness was 225.55 μm and 215.7 μm, and the mean RNFL thickness was 108.39 μm and 104.61 μm, in the amblyopic eye and the normal eye, respectively. OCT assessment of RNFL thickness and macular retinal thickness revealed a significantly thicker in amblyopic eye (P < 0.005).

Conclusion: The amblyopic process may involve the RNFL and the macula. However, further evaluation is needed.
FRIDAY 1 SEPTEMBER

Aniridia as a main sign of WAGR syndrome

Natalya Fomina, Elvira Saidasheva

North-West State Medical University named by I.I. Mechnikov,
St. Petersburg - Russia

Introduction: WAGR syndrome is a rare condition and is
named for its main features: Wilms tumor, aniridia, genitourinary
anomalies, and intellectual mental retardation. WAGR syndrome
is associated with deletions of the 11p13 area of chromosome
11, including the PAX6 and WT1 genes. In most cases
genetic changes occur spontaneously during early embryonic
development. The mother’s pregnancy and the baby’s birth
history are not unusual. The symptoms of WAGR syndrome
are usually seen after the baby is born. Aniridia may be the first
noticeable sign of WAGR syndrome.

Methods: Clinical case:

In 2013, the boy B. was born on 39 week of the 1-st pregnancy
of 31-year-old women (2660g / 49 sm). Apgar values were 7/8.
On the 4-th day of life the baby with many diagnosis (intrauterine-
growth retardation, posthypoxia, convulsive syndrome,
cryptorchidism, hypospadias, hyperbilirubinemia) was transformed
to the intensive care department of the St. Petersburg’s Neonatal
Hospital №17 of St. Nikolais.

L- form abnormal kidney was observed by ultrasound exam in
the hospital.

On the 12-th day of life the baby examined by ophthalmologist.
As usual the ophthalmological exam of newborns performs
after mydriasis with “Mydrimax”. During the investigation the
ophthalmologist supposed the partial aniridia of the both eyes.
Neonatologist didn’t answer about this defect during the exam.
Next day the diagnosis of aniridia was conformed. The present
of aniridia and genitourinary anomalies to allow suggest WAGR
syndrome. Genetic investigation conformed this condition.

Results: Aniridia was diagnosed only on the 13 day of life the
baby by ophthalmologist. It was difficult to recognize partial
aniridia after mydriasis.

Conclusion: To improve the quality of the neonatal exam may
be useful to make investigation with modern non-contact non-
mydriasis hand-held medical camera that provides high image
quality of anterior segment structure and fundus of the
newborn’s eye.

Bilateral idiopathic macular choriocapillaritis
in an otherwise healthy 11 year old boy

Catherine Cassiman, François Vermeulen, Carine Wouters,
Lien De Somer, Joachim Van Calster, Ingele Casteels

University Hospitals Leuven, Department of Ophthalmology,
Leuven - Belgium

University Hospitals Leuven, Department of Pediatrics,
Leuven - Belgium

Introduction: We present a case of an 11 year old boy, referred
to us for bilateral papilledema and anterior uveitis. The following
months he developed a large macular zone of choriocapillaritis
and consequently lost central vision in both eyes. A neovascular
membrane at the scar edge and progressive peripheral
inflammation was noticed recently after one year of immune
suppressive treatment. Clinical pictures and work up will be
presented as also an open question to the audience for further
diagnostic suggestions to discover the underlying etiology.

Methods and Results: The ophthalmological images from
presentation until present will be shown together with all relevant
clinical and technical investigations. No underlying etiology
could be confirmed. The disease was classified as an idiopathic
autoimmune choriocapillaritis and immunosuppressive therapy
with oral steroids, subcutaneous methotrexate and adalimumab
was adjusted. One year after treatment, together with tapering of
steroids, new active peripheral lesions were discovered.

Conclusion: We present a bilateral idiopathic macular and
peripheral choriocapillaritis in an, otherwise healthy 11 year old
boy. After exclusion, no definite diagnosis could be established.
The clinical picture as we present was never published before, the
underlying etiology remains unsolved.
Novel mutations of the RDH12 and MERTK gene in a family with early onset retinal dystrophy

Monika Grudzinska Pechhacker
St. Erik Eye Hospital, Stockholm - Sweden

Introduction: Retinitis pigmentosa (RP) is a clinically and genetically heterogenous group of inherited retinal dystrophies. To date, over 80 genes have been implicated in RP. However, the disease demonstrates significant locus and allelic heterogeneity not entirely captured by current testing platforms. The purpose of this study was to characterize the underlying mutation in a family, where half of the children were affected with early onset retinal dystrophy.

Methods: The phenotype was determined with help of fundus photography, SD-OCT, visual field test and electrophysiology. Oligonucleotide-selective sequencing of the affected proband was performed. Deletions and duplications were detected from targeted next-generation sequencing data. Variants classified as likely pathogenic has been tested using Sanger sequencing.

Results: Three sequence variants were identified in the proband in known RP-associated genes. Sequence analysis revealed that the proband was a homozygote in two independent missense mutations in RDH12 and MERTK genes, and heterozygote in FZD4 gene. The clinical picture suggests previously described RDH12-retinopathy.

Conclusion: Our study identifies novel mutations in a family with early onset retinal dystrophy. This study also demonstrates the clinical value of early genetic testing. Would a future treatment become available, the optimal time of intervention should be at a young age.

Accidental retinal finding can sometimes be lifesaving

Kaspars Ozols, Antra Treija, Sandra Valeiņa
Children’s Clinical University Hospital, Riga - Latvia

Purpose: To report a case of lipemia retinalis in a 5 years old boy - the first presentation of congenital severe hypertriglyceridemia.

Methods: Retrospective case review of a patient who came to a regular ophthalmological check-up suspected of having ametropia. Investigations showed that patient had significant hyperopia (+7 diopters in both eyes). Fundus examination revealed suspicious findings that suggested diagnosis - retinal vasculitis, blood vessels appeared white with hazy edges. Findings were documented by OCT and Fundus photography. Further investigation didn’t confirm the diagnosis of vasculitis, but blood analysis showed blood lipids in extremely high numbers:

- Total cholesterol: 20.10 mmol/L (2.0 - 4.4 mmol)
- HDL 0.44 mmol/l (<1.0)
- LDH 1.39 mmol/l (1.7 - 3.0)
- TG 115.95 mmol/l (0.1 - 1.7).

The findings were defined as a lipemia retinalis - a complication of congenital severe hypertriglyceridemia (type I hyperlipoproteinemia presumably caused by LPL gene mutation).

Results: The patient’s food lipid intake was restricted to 20 g per day or 15 % total calorie intake. After 3 months blood lipids became in reference range and retinal changes disappeared - documented on OCT and fundus photo.

Conclusion: Type I hypertriglyceridemia due to lipoprotein lipase deficiency occurs in approximately 1 case per 1 million and can be life threatening due to high risk of acute pancreatitis. In this case careful and comprehensive ophthalmological examination led to a very rare diagnosis which helped to escape severe complications of this pathology.
FRIDAY 1 SEPTEMBER

Case Report: An indicental finding of severe familial exudative vitreoretinopathy

Sabeera Hussain, Michael Iveson
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Introduction: A healthy 4 year old Pakistani girl was seen in our department with an indicental finding of blindness in her right eye which was initially recognised by a nursery school teacher during a pirate role play session. The young girl was wearing an eye patch and subsequently fell over during the role play. Her parents (known to be second cousins) had noticed an abnormal red reflex in photographs over the past 12 months but thought this was insignificant. There was no family history of eye disease. On examination VA in the right eye was NPL and fundus examination revealed leucocoria, extensive global retinal changes, a solid retinal detachment inferiorly with chronic changes involving the choroid. Left eye VA was 6/6 with a normal red reflex. Fundus examination of the left eye showed significant exudates nasal to the disc and temporal to the macular with macular preservation. Additionally the left eye also revealed extensive telangiectatic changes with significant exudates inferiorly. The patient was referred for urgent assessment by a Paediatric Specialist. As both eyes are affected a diagnosis of Autosomal Recessive Familial Exudative Vitreoretinopathy is suspected. The key priority of management is to preserve sight in the left eye.

Results: The child has been investigated for FEVR. A fundus fluorescein angiography and examination under anaesthetic has been performed. Laser treatment and Avastin (anti-VEGF) has been organised. Genetic counselling has also been arranged.

Conclusion: Familial exudative vitreoretinopathy (FEVR) is a rare inherited disorder of retinal angiogenesis. A severe form of the disease can lead to complete blindness therefore early recognition and management is vital to prevent disease progression. Genetic counselling and support for families also forms an integral part of care.

Situational analysis of paediatric tertiary ophthalmology facilities in african VISION 2020 LINKS

Axelle Rigaudy
London School of Hygiene and Tropical Medicine, London - UK

Introduction: The burden of childhood blindness totals 1.4 million children worldwide, with Sub-Saharan Africa disproportionately affected. Tertiary Paediatric Ophthalmology facilities are essential in tackling important causes of avoidable blindness such as childhood cataract. This study assesses the current situation in 16 African tertiary paediatric ophthalmology facilities part of the VISION 2020 LINKS programme (health partnerships between the UK and mainly African eye units) to help determine need, strengthen the eye health system, and ultimately reduce childhood blindness.

Methods: A descriptive cross-sectional survey was conducted in 16 tertiary Paediatric eye care centres (n = 16) part of the VISION 2020 LINKS programme. Data was collected using comprehensive questionnaires based on WHO health system blocks: governance, human resources, training, infrastructure, equipment, finance and service delivery. Questionnaire responses were analysed using SPSS, Microsoft Excel and according to a scoring system based on WHO recommendations for Child Eye Health Tertiary Centres (CEHTFs) and Aravind vision 2020 e-resource.

Results: Paediatric Tertiary Ophthalmology facilities will likely need significant strengthening in all areas. This may be difficult to achieve without further funding. The variation in skill levels and resources amongst centres mean increased collaboration, sharing of experiences, and support between centres may provide a possible solution to increase the capacity and quality of services by optimising available resources.

Results: The child has been investigated for FEVR. A fundus fluorescein angiography and examination under anaesthetic has been performed. Laser treatment and Avastin (anti-VEGF) has been organised. Genetic counselling has also been arranged.

Conclusion: Familial exudative vitreoretinopathy (FEVR) is a rare inherited disorder of retinal angiogenesis. A severe form of the disease can lead to complete blindness therefore early recognition and management is vital to prevent disease progression. Genetic counselling and support for families also forms an integral part of care.
Case report of acute posterior multifocal placoid pigment epimelipathiy

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Introduction: Acute posterior multifocal placoid epitheliopathy (APMPPE) is a rare, idiopathic disorder characterized by discrete areas of subretinal inflammation, and has a tendency to occur between the 2nd and 4th decades of life.

Methods: To present a case report of APMPPE in a 10-year-old boy that developed after an influenza-like illness prior to this ocular pathology in 2010 and to demonstrate a long-term visual outcome till 2017. Optical coherence tomography (OCT) was used to measure the macular thickness; fluorescent angiography (FA) was performed to confirm the diagnosis.

Results: The initial visual symptoms involved blurred vision, paracentral scotomas and multiple floaters. Visual acuity (VA) at presentation was 20/30, then decreased till 20/400 in both eyes. External and slit lamp examinations were unremarkable, and intraocular pressures were within normal limits. Fundoscopic examination of both eyes indicated numerous, flat, yellow-white placoid lesions at the level of the retinal pigment epithelium located throughout the posterior pole. Macular thickness in the right eye was 179 μm and 144 μm in the left one based on OCT measurements. FA showed bilateral multifocal hypo-fluorescent lesions with hyper-fluorescence during the late venous stages of the study, evident in both eyes several minutes following injection. The patient was treated with systemic steroids to control the course of ocular disease. VA at the last follow-up in 2017 was 20/25 in both eyes with correction of myopic astigmatism.

Conclusion: The choriotereinal scars did not enlarge with time. APMPPE has a good long-term prognosis for VA, although residual symptoms and paracentral scotomas are still present.

Optical coherence tomography angiography findings in leber congenital amaurosis

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Introduction: Leber congenital amaurosis (LCA) is the most severe and earliest form of inherited retinal dystrophies associated with severe visual impairment before age of 1 year. Fundus findings range from normal to severe retinal findings such as macular coloboma.

Methods: Optical coherence tomography angiography (OCT-A) findings of a patient that had the diagnosis of LCA and macular coloboma were reported.

Results: A 17 year old girl who was followed with the diagnosis of LCA since 6 months of age was evaluated with OCT and OCT-A. She had 4 siblings with the same diagnosis. On ophthalmological examination, best corrected visual acuities were 15/100 OD and 30/100 OS with normal anterior segment findings and nystagmus. On fundus examination, there was diffuse peripheral pigmentary retinal degeneration as well as attenuated vessels. Optic disc OCT-A showed peripapillary vascular anomalies and this was stable during her follow-up.

Conclusion: OCT-A is a recent, non-invasive method for evaluating normal vasculature and vessel anomalies. It can give more information about associated vessel anomalies in LCA.
**FRIDAY 1 SEPTEMBER**

**OCT angiography findings in Leber Hereditary Optic Neuropathy**

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**Purpose:** To report the findings detected with optic coherence tomography (OCT) and OCT Angiography (OCT-A) in follow up of a 10-year old boy that was diagnosed as Leber Hereditary Optic Neuropathy (LHON).

**Methods:** Initially the patient was seen at the age of 8, for control examination because of the family history for LHON at maternal uncle. His findings were normal except 10 PD of exophoria. Two years later he was seen with the compliant of decreased vision in the left eye and his visual acuity was finger counting at 1 meter in the left eye and color vision was impaired and right eye was normal. He was diagnosed as LHON and genetic testing confirmed the diagnosis with G11778A mutation. About 6 months later he had the complaint of decreased vision in the right eye. His visual acuity was 0.1 for right eye. He was followed with OCT and OCT-A and the findings were evaluated.

**Results:** At the acute phase, there was an increase in retinal nerve fiber layer thickness and a progressive decrease followed. Optic coherence angiography (OCT-A) showed reduced peripapillary vessel density at the acute and chronic phases.

**Conclusion:** OCT-A is a recent, non-invasive method for evaluating normal vasculature and vessel anomalies. It can give more information about associated, underlying vessel anomalies in LHON and may guide in diagnosis and treatment.

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**Analysis of haemodynamically parameters of ocular and renal arteries in children with chronic glomerulonephritis**

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**Introduction:** Glomerulonephritis is an actual problem in pediatrics and is characterized by the defeat of the vascular bed of all organs and tissues leading sometimes to severe, irreversible changes, and sometimes to death of patients.

The aim of the study was to perform a comparative analysis of ultrasound dopplerography of the organ of vision and kidneys in children with chronic glomerulonephritis.

**Methods:** 25 children with chronic glomerulonephritis (CH) from 7 to 14 years old were examined, nephrotic form was observed in all patients. The study of hemodynamics of the eyes was performed by the ophthalmodopplerography method with the study of the parameters of the orbital artery (HA), the central artery of the retina (CAC), the posterior short ciliary artery (CCCA), and the ischemia index.

**Results:** The study of renal blood flow revealed its changes in CGN, and the character of hemodynamic disorders was the same in all the patients examined. In the study of renal hemodynamics in 10 children (40%), a statistically significant decrease in the level of the distal part of the main renal artery of the DDS and an increase in the segmental and interstitial arteries (P <0.05) was found. More pronounced changes in MCC, KDS were found in children with CGN with hyperureukemia, with a long course of the disease. The study of hemodynamics of the eyes revealed a violation of blood circulation with preservation or impairment of visual functions. Thus, in children of CGN patients, the Resistance Index (RI) was reduced (P <0.05), while the Pulsational Index (PI) was elevated (P <0.05). Vmax and Vmin in the CVS, Vmax and Vmin in the CCAR and in the HA are reduced, a more pronounced significant decrease was found in children with a long course of the disease (P <0.05). RI and PI were within normal limits.

The conducted studies allow us to conclude that for children of CGN patients, nephroangiopathy and angiopathy of the retina vessels are characteristic, a decrease in the velocity parameters of venous outflow in MCC and CDS, CVS, a decrease in the velocity parameters of the blood flow along the CCVC and the orbital artery. These facts make it possible to establish the presence in patients with CGN of ischemic damage to the vessels of the kidneys, the eyes that were more pronounced in children with hyperuricemia and a prolonged course of the disease.
Asymptomatic autosomal recessive bestrophinopathy: Can it be treated?

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Introduction: Autosomal recessive bestrophinopathy (ARB) is a disorder caused by biallelic mutations in the BEST1 gene. It has a variable phenotype with extramacular multifocal yellowish subretinal deposits, extensive subretinal fluid, and cystoid macular edema. Carbonic anhydrase inhibitors have been used in the treatment of cystoid macular edema in several other retinal dystrophies such as retinitis pigmentosa and juvenile retinoschisis.

Purpose: To describe the clinical findings in an 8 years old boy with autosomal recessive bestrophinopathy treated with topical carbonic anhydrase inhibitors (CAI).

Methods: We present the case of a patient with ARB including a complete ophthalmic examination, fundus photography, spectral-domain optical coherence tomography (SD-OCT), fluorescein angiography, fundus autofluorescence imaging, full-field electroretinography (ERG) and electro-oculography (EOG).

Results: An asymptomatic 8 years old boy with 6/6 visual acuity has been found to have multiple bilateral extramacular yellowish subretinal deposits. On further investigation, SD OCT demonstrated a shallow serous retinal detachment throughout the macula, hyper-reflective subretinal deposits, elongation of photoreceptor outer segments and cystoid macular edema. Due to the presence of subretinal fluid and cystoid macular edema he has been treated with Dorzolamide three times a day. SD OCT was repeated after treatment with topical CAI.

Conclusion: We present a case of an asymptomatic 8 years old boy incidentally diagnosed with autosomal recessive bestrophinopathy. We discuss the natural history of ARB, the necessity of treatment in asymptomatic case and the result of treatment with Dorzolamide.

X-linked retinoschisis - a case report of genetically proven X-linked retinoschisis of two brothers

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Introduction: X-linked juvenile retinoschisis (XLRS) is a condition that affects mostly males and is characterised by impaired vision from early childhood. Most cases are caused by a mutation in RS1 gene, which encodes retinoschisin protein. Retinoschisin is expressed and secreted from photoreceptors and bipolar cells, and it binds strongly and specifically to the surfaces of many cells in the retina. Mutated forms of retinoschisin are unable to bind properly, which leads to splitting (schisis) of the retinal layers.

Methods: The case reports of two brothers of a family of 6 children (4 boys and 2 girls) are described. The two patients have been followed by the author since August 2016. Both patients underwent visual acuity testing, cycloplegic refraction, fundus imaging and OCT after which the diagnosis of XLRS was suspected.

Results: The two male patients (6 and 3 years old) referred to an ophthalmologist since their mother had noticed her two sons had bad vision. Both patients were diagnosed with myopia and prescribed glasses. However, visual acuity did not improve after correction. Macular pathology was found with ophthalmoscopic examination and OCT imaging revealed macular retinoschisis of both patients. The older patient underwent ERG testing, which confirmed the diagnosis of XLRS. The older patient underwent genetic testing and a hemizygous mutation in RS1 gene c.637C>T (p.Arg213Trp ) was detected. The other two brothers and two sisters of the family have also been examined and no pathology was found.

Conclusion: Here I present a case of two brothers with XLRS. It is important to recognise the pathology and be aware of the risks of the disease. Family counselling is important to inform them about the inheritance pattern. Affected patients are recommended to avoid activities such as contact sport, which may pose an increased risk for retinal detachment. At the moment there is no treatment available for the disease, but lately gene therapy has been shown to be a promising option for patients with XLRS.
Toying with the epiCam: enhancing the view in infants
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Introduction: Imaging of the paediatric fundus is challenging with handheld devices but, in the case of ROP screening, may reduce the frequency of attendance of an ophthalmologist, e.g. in remote areas or busy units. Cameras may be operated by trained nursing staff, enabling telemedicine approaches as reported in several Retcam studies. However, the cost of this device is prohibitive for many hospitals, even in developed countries. The advent of low-cost high-tech imaging modalities should eventually solve this problem. Currently, there are no low-cost wide-field cameras on the market. Recently, the epiCam was introduced, initially as a black and white camera and later as a colour video camera. The camera provides a 33° vertical x 45° horizontal field through a dilated pupil allowing posterior pole images in co-operative patients.

Methods: To explore the operability of the epiCam in young infants and children epiCam recordings were made during routine ROP-rounds and other examinations in children (sometimes sedated) in an attempt to provide more objective recordings than written findings. The epiCam M and C were employed using eye lid specula and/or fundus contact lenses. Subsequently photographs and video stills were judged for clarity and retinal detail (graded poor, moderate and good), extent of field of view (zone 1, zone 2) and whether the image would allow a judgement regarding the presence of plus disease where applicable.

Results: 10 premature babies and 7 children were examined with the epiCam M and 5 premature babies and 7 children with the epiCam C (age range 30 weeks GA to 15 years). 27 right eye images were evaluated. Posterior pole images could be obtained in all children, even with ventilatory equipment in place. Image quality was graded poor in 3 cases, moderate in 19 and good in 5. In 17 imaging was greatly enhanced by the application of a Koeppe-style contact lens. For a view into zone 2 the contact lens was essential. In addition focusing was easier to allow judgement of plus signs (where applicable) in 10 out of 14.

Conclusion: The epiCam allows posterior pole imaging of the paediatric fundus. In premature babies, young infants and sedated children the field of view is greatly enhanced by placing a Koeppe-style contact lens. This suggests that further adaptation of the epiCam’s housing and optical components might provide a suitable low-cost wide-field imaging module for infants in the future.

Foveal hypoplasia: normal does not always mean normal and abnormal is not always abnormal
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Introduction: Foveal hypoplasia, defined as an undeveloped fovea, is usually described in associations with other ocular disorder but could be as an isolated entity.

Purpose: To present the clinical variation of foveal hypoplasia.

Methods: We describe 3 presenting cases of the variation of foveal hypoplasia. The clinical finding and the OCT results of the 3 cases and their first degree family members are discussed.

Results: Case 1- 6 year old boy present with decreased vision. Ophthalmologic evaluation revealed no abnormality. OCT showed foveal hypoplasia grade 4. His father’s OCT showed foveal hypoplasia grade 1.
Case 2- 10 year old boy with good visual acuity was followed for esotropia. Ophthalmoscopy showed irregular macular reflex. OCT revealed foveal hypoplasia grade 4. His brother was found to be albinotic.
Case 3- 6 year old girl with excellent visual acuity was referred for the evaluation of a funny-looking fovea in both eyes. OCT revealed foveal hypoplasia grade 4. On OCT angiography there was no evidence of foveal avascular zone in the superficial and deep retinal capillary plexus. All family members and their OCT were normal.

Conclusion: It is already a common practice to perform OCT in any case of unexplained decreased visual acuity. High grade foveal hypoplasia can be found in OCT even in the presence of good visual acuity. Evaluation of family members can help in distinguish between familial and sporadic foveal hypoplasia even in the presence of different phenotypic expression. A none familial case of foveal hypoplasia, even if high grade may have favorable visual acuity result.
Novel secretome analysis to identify crucial signaling proteins in the cellular communication

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Introduction: As the eye is integrated into the human organism with its different organs and numerous cell types, the intercellular communication plays a key role in its development and other relevant biological processes such as healing and immune response. One innovative and novel way to investigate the protein-based communication of cells are secretome analyses, which provide a comprehensive study of proteins that are secreted by a cell, tissue, organ or organism at any given time and under certain conditions.

Methods and Results: Given the relevance of cellular communication in general, we established a novel setup to investigate the secretome of different cells such as primary cardiomyocytes and the protein-based interaction with macrophages. The combination of pulsed stable isotope-labeled amino acids (SILAC) for reliable quantification and azidohomoalanine (AHA), an azide-bearing analogue of methionine, for the enrichment of newly synthesized and secreted proteins, allowed the analysis of primary cells without starvation medium. This approach enabled the detection of newly synthesized proteins with low abundance in a background of high abundance of serum proteins. In comparison to previously published analyses this significantly increased the sensitivity and yielded in more than a 2-fold increase in the measurement of secreted proteins. Validation experiments in cell culture and in mice confirmed the upregulation of previously unknown proteins. To further investigate cell-cell-communication on the protein level, we performed co-culture experiments of cardiomyocytes and macrophages under various conditions. This setup identified established and previously unknown pro- and anti-inflammatory signaling molecules as well as biomarkers of ischemia that are currently being validated and further investigated.

Conclusion: We here describe a novel multidisciplinary method to detect the secretome of different cell types under ischemic and other stress conditions as well as in the presence of inflammatory cells. Validation experiments confirmed the translatable into the context of in vivo processes and hint that this approach can lead to the discovery of crucial signaling proteins in the pathophysiology of different organs. Therefore, this approach has the potential to discover unknown disease specific pathways and signaling proteins involved in the intercellular communication of the eye.

Case report: Hypertensive retinopathy

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Introduction: Hypertension is associated with cardiovascular risk and systemic target organ damage. Retinopathy is considered one of the indicators of target organ damage. Systemic arterial hypertension in children has been thought to be secondary in origin. The most common etiology leading to hypertension in children is a renal disease but hypertension itself can result in renal injury and failure secondary to loss of autoregulation of renal blood flow.

Methods: A clinical case study of renal disease ophthalmic manifestations and Dynamics of RNFL, macula thickness and visual acuity.

Results: 11 year old girl came for regular ophthalmological examination and complained about decreased vision in the left eye (V od=1.0; V os=0.2). Fundus examination showed bilateral optic disc edema, macular edema and macular exudates (macular star). After multiple blood pressure measurements the girl was hospitalized in child hospital for more detailed general examination. When additional anamnesis was asked-girl had no significant complains about headaches or weakness. Renal scintigraphy, ultrasound of kidney and laboratory data revealed decreased, nonfunctional right kidney. After right nephrectomy visual acuity in the left eye improved to 0.7, optic disk edema and macular edema decreased. Blood pressure was limited in acceptable range using amlopidini and fosinoprili.

Conclusion: It is important to remember that regular ophthalmological examination can turn out to be an emergency situation. Timely patients health conditions assessment and multidisciplinary approach can lead to good results.
**FRIDAY 1 SEPTEMBER**

**Different phenotype in three siblings with a mutation in the ABCA4 gene – a case report**

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**Introduction:** The aim is to report optical coherence tomography (OCT) and electroretinogram (ERG) findings in three siblings with a mutation in the ABCA4 gene.

**Methods:** Three girl siblings were referred for ERG to our department due to visual loss. All three sisters were assessed with visual acuity (VA) and colour vision testing, perimetry, OCT, autofluorescence, ophthalmoscopy, fullfield (ffERG) and multifocal ERG (mfERG). Blood samples for genetic analyses were taken.

**Results:** The girls were 9, 8 and 6 years of age at examination. The VA was 0.03/0.02, 0.1/0.1 and 0.5/0.5 decimal Snellen acuity, respectively. They had similar ffERGs and mfERGs. In particular, the mfERG was reduced, but the 30 Hz flicker of the ffERG was also affected. The OCT images were similar in the two children with the severely reduced VA. The retina was thin and the photoreceptor layer of the fovea disappeared. In the child with the better VA, OCT revealed an intact photoreceptor layer and a shallow fovea with a thickening of the inner retinal layers. Gene analyses showed a homozygote mutation in the ABCA4 gene (c.5917delG).

**Conclusion:** Mutations in the ABCA4 gene are associated with a spectrum of retinal dysfunctions, not only Stargardt disease. In these sisters, the cone system of the ffERG was also affected, suggesting a more generalized disease. The OCT finding was different in the sister with the better VA, which might represent an early stage of the disease or a different phenotype. Follow-up of the ERG and OCT is planned.

**SATURDAY 2 SEPTEMBER**

**Rare cause of bilateral optic disc oedema in a 2-year old girl**

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**Introduction:** We present a 2-year old girl with bilateral optic disc oedema.

**Methods:** Case report

**Results:** Now almost 3-year old girl presented in the neonatal period with skin rash and fever. Brain MRI showed subtle subcortical nonmyelinated regions. Due to low levels of virus detected in her blood congenital CMV infection was suspected. She was sent for an ophthalmology assessment.

Visual acuity (VA) at 3 months was 6/190 (preferential looking test -PL), and there were no signs of intraocular inflammation or optic disc involvement. Six months later her VA was 6/130 PL, hypermetropia was found and corrected. No other clinical changes were noted and electrodiagnostic testing (VEP, ERG) was normal.

Fever resolved spontaneously whereas bursts of skin rash persisted. At the age of 12 months she was assessed by clinical immunologist due to recurrent skin rash, persistent CMV viral load and elevated inflammatory markers. Immunodeficiency was suspected and she was sent for ophthalmology reevaluation.

At the age of 18 months bilateral optic disc oedema without haemorrhages was found. Ultrasonography excluded optic disc drusen. VEP was normal. Repeated brain MRI was suspicious for raised intracranial pressure and subcortical lesions of leukomalacia. Spinal fluid evaluation was negative for CMV infection, did not show signs of inflammation and could not confirm elevated intracranial pressure. Optic disc oedema persisted despite oral acetazolamide therapy.

When periodic fevers reappeared auto-inflammatory syndrome with CNS involvement was suspected. Genetic testing confirmed mutation in NLRP3 gene that causes Chronic Infantile Neurological, Cutaneous and Articular Syndrome/Neonatal Onset Multisystem Inflammatory Disease (CINCA/NOMID). After treatment with IL1 blockade (Anakinra) periodic fevers, rashes and inflammatory markers resolved within days. Optic disc oedema improved within weeks. Her best corrected VA at last follow up visit was 0,7 Snellen chart (Lea optotype).

**Conclusion:** We present a girl with bilateral optic disc oedema caused by an auto-inflammatory CINCA/NOMID syndrome. This syndrome is characterized by skin rash, joint manifestations and involvement of CNS including sensory organs. Almost 83% of patients have optic disc involvement (optic disc oedema, papilloedema or optic atrophy) and one-fourth of untreated patients can develop moderate to severe visual loss. With appropriate and early treatment we hope to prevent significant visual loss and blindness.
Bilateral inferior corneal pseudopterygium in Kabuki syndrome

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Wolfson Medical Center, Holon - Israel

Introduction: Kabuki (Niikawa-Kuroki) syndrome is a rare multi-organ syndrome characterized by a characteristic peculiar face, skeletal anomalies, dermatoglyphic abnormalities, varying degrees of mental retardation and growth deficiency. The most common ocular features described are strabismus and ptosis. Many other ocular features have been previously described, including: amblyopia, nystagmus, microcornea, blue sclera, cataracts, nasolacrimal duct obstruction, corneal pannus and more.

Purpose: To describe an additional ocular feature in children with Kabuki make-up syndrome: a bilateral inferior corneal pseudopterygium.

Methods: A case series analysis of eight patients with sporadic Kabuki syndrome.

Results: In four of eight children with sporadic Kabuki syndrome, an inferior corneal pseudopterygium was observed in both eyes. In all affected children the eyes were exposed during sleep. In one girl, the fibrovascular membrane encroached toward the visual axis in one eye. The membrane was excised and recurred twice.

Conclusion: Corneal inferior pseudopterygium and exposure during sleep may be associated with Kabuki syndrome.

Ophthalmic findings in pediatric patients with chronic autoimmune thyroiditis

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Introduction: Chronic autoimmune thyroiditis is the most common cause of acquired hypothyroidism in children and adolescents. Among many symptoms of hypothyroidism eye abnormalities can be also noticed in children, mostly mild inflammatory symptoms, but in some cases proptosis and eye motility deficits occur.

Methods: The aim of this study was to evaluate the prevalence, type and risk factors of developing ocular abnormalities in pediatric patients with chronic autoimmune thyroiditis. Enrolled patients included 38 children aged 7-18 years old and control group comprised of 48 healthy children aged 5-18 years old. Full ophthalmological examination, Schirmer test and tear break up time test (TBUT) were performed.

Results: Different ocular signs were reported by 60.5% children with chronic autoimmune thyroiditis. None of the patients demonstrated proptosis or eye muscles restriction, lower eyelid retraction was diagnosed in 5.3% of cases. Intraocular pressure was within normal range in all children, but mean intraocular pressure values were higher in children with chronic autoimmune thyroiditis than in control group. Inflammatory symptoms (mostly redness of the eyelid and conjunctiva) were present in 31.6% children with chronic autoimmune thyroiditis. One third of patients exposed to tobacco smoke presented inflammatory symptoms. Results of Schirmer test and TBUT were abnormal in 25% and 57.9% cases respectively.

Conclusion: This study has revealed that pediatric patients with chronic autoimmune thyroiditis may present ocular abnormalities. Awareness of pathologies such as inflammatory symptoms of soft tissues and anterior segment and dry eye syndrome in children with chronic autoimmune thyroiditis might be helpful in assessing these patients in clinical practice.
**Sustained effect of somatostatin analogue treatment on visual recovery in a 13-year-old boy with advanced thyrotrophic pituitary macroadenoma**

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Introduction: Pituitary adenomas are rare tumours accounting for 2.7% of supratentorial tumours in childhood. Early ophthalmology input is required due to visual field defects and impaired visual acuity caused by vision-threatening compression. When surgical debulking is not possible, medical therapy with octreotide, a somatostatin analogue has been used. We present a rare case of a child with thyrotropic macroadenoma responding to octreotide.

Methods: The ophthalmic case notes for a 13-year-old boy were reviewed, retrospectively. All data from his ophthalmic assessments were evaluated by a masked observer to see if there was any correlation between visual recovery and timing of somatostatin analogue treatment.

Results: A 13-year-old male patient presented with a one year history of headache and reduced visual acuity. Presenting visual acuity was 0.1 logMAR on the left and 0.8 logMAR on the right with a relative afferent pupillary defect (RAPD). Initial Goldmann perimetry revealed a left temporal hemianopia and extensive right central and paracentral visual field loss. He also had pale optic discs and ishihara colour plates of 16/17 on the left and 3/17 on the right. Magnetic resonance imaging and subsequent biopsy confirmed a thyrotropic pituitary macroadenoma. Unfortunately, post-operative recovery was complicated with a three week stay in intensive care. Further debulking neurosurgery was decided against by the parents and surgeon due to the previous complications. Instead, a trial of somatostatin analogue treatment was given resulting in rapid recovery of his visual acuity and fields in both eyes. Discontinuation of treatment, caused by non-compliance, resulted in deterioration in visual function. This recovered after re-treatment with a visual acuity of 0.0 logMAR in the left and 0.2logMAR in the right with full colour vision and no RAPD. Remarkably, his visual field in the left was full and on the right had only mild paracentral loss.

Discussion: We report significant visual recovery in a child with advanced thyrotrophic pituitary macroadenoma-related visual pathway compression in response to somatostatin analogue therapy. A cause-effect relationship is further supported by the transient deterioration in visual function during temporary cessation of treatment and subsequent improvement on retreatment. We report a follow-up of 27 months with no deterioration due to tolerance or resistance during somatostatin analogue treatment so far.

**Visual perceptual disabilities in children adopted from Eastern Europe**

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Introduction: Children adopted from Eastern Europe often show neurodevelopmental, behavioural, social and emotional disorders. Visual perceptual and visual motor skills seem to be a good way to assess cognitive domains in these children.

Methods: We compared two cohorts of infants aged between 5 and 18 years. Seventy-nine boys and girls, adopted from Eastern Europe, were included in the study group. The control group was formed by age and gender matched children born in Spain. All children underwent a full ophthalmologic assessment and standardized testing of visual cognitive skills (Test of Visual Perceptual Skills, Test of Visual Analysis Skills and Facial Memory subtest from the Test of Memory and Learning).

Results: Adoptees presented worse visual motor and visual perceptual outcomes in all skills (visual discrimination, visual memory, spatial relationships, form constancy, sequential memory, visual figure-ground and visual closure) compared with control subjects, with statistically significant difference in TVPS global centile (66.8 vs 50.3; p=0.001), spatial relationships skill (81.9 vs 64.6; p=0.004) and visual figure-ground skill (74.1 vs 52.1; p=0.002). Face recognition was as well significantly lower in adopted children (57.1 vs 42.4; p=0.009). Twenty one adopted children (26.6%) had sentinel finding for foetal alcohol spectrum disorders (FASD). Main facial features relates to FASD correlated to visual cognitive skills outcomes found. Of the adopted children, those diagnosed of FASD showed an incrementally worse visual perceptual and visual motor outcomes (TVPS global centile=36.86; p=0.001; TVAS=10.38; p=0.002).

Conclusion: Children adopted from Eastern Europe are at increased risk of visual perceptual disabilities, especially those with foetal alcohol spectrum disorders.
**A quantitative study of fixation stability in amblyopia**

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**Introduction:** To analyze fixation stability in children with amblyopia and determine whether fixation instability is correlated with the magnitude of visual acuity deficit.

**Methods:** Twenty-five children (4-10 years old) with strabismus, anisometropia or both conditions were included in the study: 12 with varying severity of amblyopia and 13 aged-matched nonamblyopic subjects. Fixation stability was measured using a digital device with remote eye tracking technology incorporated, which records eye position at 60 Hz. Quantitative parameters were calculated to characterize fixation: saccadic reaction times (SRT) and stability of fixation. Fixation stability was quantified as the 68% bivariate contour ellipse area (BCEA), the best-fit ellipse within which 68% of fixations occurred during the presentation of several 4-second visual stimuli.

**Results:** Children with amblyopia had larger BCEAs for amblyopic eyes (mean=7.64 deg²) than fellow eyes (mean=4.62 deg²) and right eyes of the nonamblyopic subjects (mean=4.23 deg²), with longer SRTs (0.28 and 0.21 seg for amblyopic and fellow eyes, respectively) in children with amblyopia compared with nonamblyopic group (0.17 seg) and with a group of healthy controls (0.14 seg).

**Conclusion:** Amblyopic eyes with poorer visual acuity had greater fixation instability. Gaze stability disruptions could be considered as prognostic factors in amblyopia.

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**Corneal microcysts due to Cytarabine toxicity**

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Department of Ophthalmology, University Hospital Ghent - Belgium

**Purpose:** Case report

**Results:** A nine-year old girl with Acute Lymphocytic Leukemia (ALL), complained of ocular itching, burning and photophobia in both eyes two days after the cessation of the intravenous chemotherapy with Cytarabine. Bilateral corneal epithelial microcysts, more densely distributed near the center of the cornea than in the midperiphery, were noted by slit lamp. Topical treatment with Dexamethasone Monofree three-hourly was started. The cysts resolved completely after three days. The steroid drops were tapered down and could be discontinued after nine days.

**Conclusion:** Cytarabine is an antimetabolite and interferes with DNA synthesis by blocking the function of DNA polymerase. This chemotherapeutic is known to penetrate body fluids, including crossing the blood-brain barrier. It can also be found in the aqueous and tears. Corneal toxicity occurs after five-to-seven days of treatment. The transient amplifying cells, located in the basal cell layer of the corneal epithelium are more susceptible to the Cytarabine toxicity than the epithelial stem cells. Although the microcysts do resolve spontaneously, a more favourable outcome has been reported with the use of topical steroids. A prophylactic treatment with topical steroids can be recommended. The mode of action, by which topical steroids prevents the formation and hastens the resolution of microcysts, is not known.
Causes of congenital optic nerve abnormalities

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Introduction: The formation of the optic disc and optic nerve fibers occurs between 3 and 10 weeks of gestation. Violations of the processes of embryogenesis and differentiation during this period lead to anomalies in the development of the optic nerve. Genetic changes cause blindness in children in 50% of cases. Unfortunately, etiology remains unknown in more than 50% of cases.

Purpose: To study the etiological factors leading to congenital anomalies of the optic disc.

Methods: To diagnose congenital anomalies of the optic nerve disk were used anamnestic data, ophthalmological examination methods of 27 children.

Results: Disturbances in the process of closure of the embryonic cleft lead to coloboma, disruption of the replacement of ganglion cells by fibers - to hypo- or aplasia of the optic nerve, disturbances in the involution of hyaloid vessels - to the persistence of the primary vitreous body. The interaction of a large number of genes, many of whose products are transcription factors, plays an important role in regulating the complex mechanisms of proliferation, differentiation and death of cells occurring in the process of bookmarking and developing the organ of vision. These are: PAX6, TGFbeta family, FOXC1, FOXC2, FOXE3, PITX2, PITX3, LMX1B, CYP1B1. Disturbances in the structure of these genes or their interactions lead to gross violations of development. For heterozygotes, mutations in the genes SOX2, BMP4, PAX6 and a number of others are characterized by a pronounced dysgenesis of the eye structures.

Conclusion: It is important to carefully specify all possible effects of damaging factors during pregnancy, and also to study the features of the clinical manifestations of various genetic variants of congenital ophthalmopathology and conduct clinical-genetic correlations that allow optimizing the DNA diagnostics process aimed at finding mutations in a particular gene.

Ophthalmologists remember Rickets: Nutritional Vitamin D Deficiency causing severe visual impairment in a child

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Homerton University Hospital, London - UK
Great Ormond Street Hospital, London - UK

Introduction: There is an increasing recognition of the prevalence of Vitamin D deficiency in adults and children, and its clinical implications. We report the first case of severe visual impairment, in a child, due to nutritional Vitamin D deficient Rickets.

Methods: We present the case of a 4-year-old child who presented with optic atrophy secondary to craniosynostosis following previously treated rickets. At presentation, his visual acuity was noted to be 1.30 in both eyes. The pupilary responses were normal, but dilated fundoscopy revealed bilateral optic nerve pallor. At the age of 10 months, nutritional Vitamin D deficient rickets had been diagnosed biochemically and radiologically, and this was treated with high dose Vitamin D. On presentation to our institution at 4 years of age, blood testing revealed insufficient serum Vitamin D, and cranial MRI showed multi-sutural craniosynostosis. Visual evoked potentials indicated marked dysfunction of the visual pathways.

Result: After treatment with Vitamin D and calvarial vault expansion surgery, six months post-operatively the visual acuity slightly improved at 0.95 in the right, and 0.9 in left eye.

Conclusion: We recommend that Vitamin D levels should be measured in all children presenting with optic atrophy and considered in cases of idiopathic papilloedema, to identify an avoidable disease process with multiple complications. We suggest that all children presenting with rickets, especially within the first 3 years of life, have ophthalmological review with dilated fundoscopy. Following this, joint ophthalmological surveillance and paediatric vigilance may be required for many years after apparent resolution of bony illness, particularly in those children with macrocephaly.
Non-surgical interventions for nystagmus developing in the first year of life (infantile nystagmus)

Rushmia Karim, Maria Theodorou
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Introduction: Nystagmus is an involuntary constant oscillatory movement of the eyes. This can be an isolated condition (idiopathic) or can occur as part of eye and central nervous system disorders.

Methods: We searched the Cochrane Central Register of Controlled Trials (CENTRAL) ClinicalTrials.gov (www.clinicaltrials.gov) and the WHO International Clinical Trials Registry Platform (ICTRP) (www.who.int/ictrp/search/en). There weren’t any date or language restrictions in the electronic searches for trials.

Selection criteria
We included randomised controlled trials (RCTs) where non-surgical interventions were used to treat infants/children/adults with nystagmus onset in their first year of life. The non-surgical Intervention could be compared with no treatment or another non-surgical intervention.

Data collection and analysis
Two review authors independently screened the search results, assessed trial quality, and extracted data using standard methodological procedures expected by Cochrane. We graded the certainty of the evidence using GRADE.

The primary outcome was change in best corrected binocular distance visual acuity.

Results: We included two trials: One randomised control trial in the UK comparing Intermittent Photic Stimulation with After Image Feedback with a control therapy in 38 patients with infantile nystagmus (7 had associated albinism); and one randomised controlled cross-over trial from the UK comparing soft contact lens (SCL) and rigid gas-permeable lenses (RGPL) in 24 participants with infantile nystagmus. In the former study there was no significant improvement in visual acuity with the treatment offered, although there was a borderline improvement in contrast sensitivity compared to the control therapy.

In the latter study there were no significant differences found in any of the nystagmus waveform characteristics assessed (intensity, amplitude, frequency, NAFX (eXpanded Nystagmus Acuity Function), longest foveation domain) between SCL, RGPL and spectacle wear. The best corrected visual acuity, reading acuity and critical print size were significantly worse for SCL wear compared to either GPCL or spectacle wear.

Conclusion: Due to the small size of the trials, there is not enough evidence to confirm/dispute the value of non surgical treatments in IN. The evidence for efficacy and safety of non surgical.

Paediatric vernal keratoconjunctivitis impact of on quality of life using Quick Questionnaire

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Introduction: Vernal keratoconjunctivitis (VKC) can affect the quality of life of patients. The paediatric population may particularly be affected with symptoms, difficult treatment regiments, frequent hospital visits and missed school days as well as limitations in desired activities. This study aims to gain an understanding of the incidence, demographics and effect on quality of life for paediatric patients affected by VKC to enable future planning of target services and management options.

Methods: The QUICK questionaire was used which has been validated for use in children with allergic keratoconjunctivitis. Questionnaires were handed during clinic consultations and vision assessment to patients with documented vernal keratoconjunctivitis. Notes were scrutinised prior to clinic and questionnairattached to front sheet of notes. Documented diagnosis of Vernal Keratoconjunctivitis by a Corneal Ophthalmologist was a requirement in the medical notes.

Results: From January 1st to March 31st 2017 40 questionaires were completed. Mean age of patients were 11 years. 70% were boys and 30% girls. Asian patients made up 35% of the group. Mean visual acuity was 0.07logMAR. 80% of patient were currently on sterois, 67.5% on allergy treatments, 27.5% on lubricants and 47.5% of patients were on both steroids and cyclosporin. No patients were on zero medication. Quality of life data is summarised in table format. Between 10-27% of patients had trouble meeting with friends, playing outdoors, practicing sport and swimming. Only 2.6% of children reported this happened all the time. Symptoms of tearing, tissue use, photophobia and red eyes were significant.

Conclusion: The study highlights the impact on quality of life facing children with VKC.
A bubble in the eye?
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Introduction: A 15-months old child was referred to our department by the Belgian screening health services because of abnormal examination with the Plusoptix Vision screenner. Parents did not notice anything abnormal with their child’s vision.

Methods: Clinical ophthalmological, orthoptic, and general examination were performed. An examination under general anaesthesia was planned.

Results: The child underwent an ophthalmological examination at the department of Ophthalmology, University Hospitals Leuven. The child had a normal following reaction to object and light, and did not resist occlusion of any eye. Biomicroscopy and eye pressure were normal; eyes were straight. The child’s general health was unremarkable. Funduscopy showed a small, well defined mass next to the optic disc in the left eye. On ultrasound, the examination, the mass showed a hyperechogenic wall and hypoechochogenic centre. Fundoscopic findings of the right eye were normal. Pictures of the fundus of both eyes were taken.

Cycloplegic refraction showed an anisometropia. The right eye was +2.5D hypermetropic. The left eye was -6D myopic. Occlusion therapy and glasses were prescribed.

An examination under general anaesthesia confirmed the presumed diagnosis of a benign vitreous cyste.

Conclusion: A 15-month old child was referred because of abnormal routine eye screening and was diagnosed with the rare but benign finding of a congenital vitreous cyst. The child had a concomitant high myopia that warrants further follow-up to prevent amblyopia.

The pathogenesis of a vitreous cyst is not clear. In this particular case, the cyst is most likely a remnant of primary hyaloid system. Another theory is that they may originate from the retinal pigment epithelium.

Vitreous cysts are in general asymptomatic and do not require any treatment.

Long term ophthalmic follow up in a case with long-chain 3-hydroxyacyl-coa dehydrogenase (LCHAD) deficiency
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Introduction: Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency is an autosomal recessive disorder of mitochondrial fatty acid beta oxidation associated with myopathy, cardiomyopathy, hypoglycemia, neuropathy and retinal changes.

Purpose: To present the course of retinal findings in a case with LCHAD deficiency.

Methods: The case history of a 6-year-old girl with LCHAD deficiency, who was diagnosed at birth and under dietary treatment, is reported. Clinical, OCT and ERG findings along the follow up are presented.

Results: In our case, a diagnosis of LCHAD deficiency was done at birth and hence early treated and followed. The girl had eye exams from the age of 1 which were normal.

At the age of 3, after systemic deteriorations, nyctalopia appeared with pigmentary retinopathy changes in both eyes. ERG was subnormal while Infra red reflectance imaging with OCT displayed more advanced stage of disease. Progressive chorioretinopathy with visual impairment was observed along the follow up on clinical exams, as well as on repeated OCTs and ERGs.

Conclusion: Retinal dystrophy in children can be related to metabolic disorders, including LCHAD deficiency. The retinal findings may rapidly progress despite dietary treatment, in relation to frequent metabolic decompensations. On early phases of the disease Infra red reflectance imaging with OCT may better identify the severity of the chorioretinopathy than ERG. Regular follow-up including OCT is recommended in LCHAD patients to monitor the ocular status.
Optic Neuropathy in Children: a case series

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Introduction: Optic neuropathy in children is difficult to diagnosis and manage. The purpose of this study is to demonstrate several cases of optic neuropathy in children with different clinical features and different result.

Methods: Case 1: A 6 yo boy was consulted from Pediatric with severe headache and diplopia since 5 days. There was history of mumps infection. There was no history of drug consumption. Position of the globe was 15 degree esotrophia. There was bilateral VIth nerve paresis. VA RE was 6/6 and LE was 6/12 uncorrected. The pupils were normally reactive to light and no RAPD. There were indistinct edge optic nerve, cup disc ratio hard to be evaluated on both eye. Brain MRI was normal. Patient was diagnosed as papilledema suspicious of idiopathic intracranial hypertension. Patient was planned to undergo lumbar puncture and got acetazolamide 3x 250 mg and potassium 300 mg. After two months, there was no headache. Position of the globe was ortophoria.

Case 2: Boy 11 yo come with sudden blurred vision 2 days before on both eyes. No history of febrile before. AVRE hand movement, AVLE 1/60, RAPD positif on right eye. Optic nerve within normal limit. MRI showed active focal area and diffusion restriction, subcortical and periventricular multiple plaque in both hemisphere and also plaque in both optic nerve. The patient was given iv steroid. After one month VA become 6/6 and 6/7.5.

Case 3: boy 14 yo come with blurred vision on LE since one month and RE since 6 month. He had history of dengue fever 6.5 month ago. There was also complaining of headache, nausea and vomiting. VA was 3/60 on the RE and 6/12 on LE. Brain MRI was normal. The patient was given iv steroid. After one month the VA become 3/60 and 6/7.5. After one year The VA become 6/6.

Conclusion: Pediatric case of optic neuropathy such as IIH or optic neuritis can be associated with previous viral infection and also multiple sclerosis. Prompt diagnosis and medical management are important to have complete resolution visual abnormalities.

Color vision testing in young children: performance comparison of normal and impaired vision subjects

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Introduction: We evaluated pseudoisochromatic plate-based color vision tests (Ishihara and Matsubara), the Mollon-Refin-Minimalist (MRM) color vision test and the Cambridge Color Test (CCT) to determine how successful young children with reduced visual acuity compared to children without visual impairment can perform the task.

Methods: A total of 23 children with reduced visual acuity (mean logMAR 0.19) and 37 children with age-related normal visual acuity (mean logMAR -0.01) aged 3-10 years were included in a prospective study. The pseudoisochromatic tests use plates, which contain a pattern (number or shape) defined by a distinct stain within an assembly of color and size randomized dots. For the MRM test, participants have to identify colored caps for the protan, deutan and tritan confusion axes with 5 different saturations each, against gray scale colored control caps. The Cambridge Color Test is a computer assisted test where the participants have to denote the direction of a Landolt C on static screens resembling pseudoisochromatic plates. The difference in chromaticity between target and background is adjusted dynamically according to the subject’s performance.

Results: All children successfully completed the Pseudoisochromatic and MRM test, CI[87%,100%] for patients and CI[91%,100%] for probands. The success rate for the CCT was 89%, CI[71%,98%] for the patients and 89%, CI[75%,97%] for the control group. The success rate for CCT varied with age group, 71% (patients) and 67% (probands) in the 3-5y olds, 92% (both patients and probands) in the 5-7y olds and 100% (both) in the 7-10y olds. The proband’s and patient’s rate of correct answers in the Ishihara test was 95%, for the Matsubara test it was 93% for probands and 99% for patients. For the MRM, 95% (versus 92%) of probands (patients) scored P1, 84% (vs. 77%) scored D1 and 91% (vs.91%) scored T1. Only one proband scored below P3/D3, most probably due to deuteranopia. Mean discrimination levels for the protan/deutan/tritan confusion axes in the CCT trivector test were 168/169/231, CI[125,211]/ [118,221]/[136,326] for patients and 117/116/137, CI [99,134]/ [95,137]/[121,153] for probands.

Conclusion: The CCT test can be performed by children with reduced visual acuity and could be useful for detecting acquired color vision defects and as an outcome measure. Reduced success rate occurred in the subgroup of 3-5y old children due to handling of the CCT equipment and limited attentiveness.
The Impact of Optic Nerve Disorders on Sleep Wake

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Introduction: Sleep deprivation increases risk of cardiovascular and metabolic disorders, cancer, and accidental injury. Disordered sleep has social and psychiatric consequences. Sleep is mediated by photic input from photosensitive retinal ganglion cells (pRGCs) via the retinohypothalamic tract to the suprachiasmatic nuclei (SCN). The SCN acts as an endogenous circadian pacemaker which is adjusted by melatonin secreted by pineal and optic tissues. Previous studies have shown increased circadian dysfunction in ocular disorders and blindness. No large-scale studies demonstrating the impact of optic nerve disorders on sleep have been performed.

We hypothesise that damage to the optic nerve affects pRGC input to the suprachiasmatic nuclei, which leads to disturbance of circadian rhythm.

Methods: 81 participants with optic nerve disorders were recruited from ophthalmology and neurology clinics at 3 UK sites. They were compared with 236 control participants recruited from 5 UK sites. Participants were recruited from September 2014 to April 2017, as part of the SOMNUS (“Effect of Ocular Diseases on Sleep and Circadian Rhythms”) Portfolio Study.

All participants completed subjective sleep quality (Pittsburgh Sleep Quality Index (PSQI)) questionnaires. In addition, 76 of the optic nerve participants and all 236 control participants completed the Hospital Anxiety and Depression Scale (HADS). Best visual acuity (of the better eye) was also recorded.

Results:
- 62.96% of participants with an optic nerve disorder had a mean PSQI>5, compared to 32.63% of controls
- PSQI was significantly worse in the optic nerve group (mean 7.54) compared to controls (mean 4.7) (P<0.0001)
- HADS Depression subscore was significantly higher in optic nerve participants (p<0.0001)
- No significant relationship was found between best visual acuity and PSQI.

Conclusion:
- PSQI was significantly worse in optic nerve participants compared to controls
- Optic nerve disease is frequently associated with chronic disorders, which may contribute to depressive mood and poor sleep
- Animal studies have shown selective preservation of pRGCs following optic nerve transection
- Patients with disrupted sleep may benefit from melatonin.

Preterm birth affects the optic nerve morphology in school-aged children

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Introduction: Premature birth affects the eyes also when the children grow up. Changes of the optic nerve head (ONH) in former preterms have been seen when analysing fundus photographs of children taken at school-age. The aim of this study was to assess the ONH morphology with Heidelberg Retinal Tomograph (HRT) and compare with children born at term.

Methods: Children, 5-16 years old, were examined with HRT assessing among other variables, rim area (RA), disc area (DA) and cup area (CA). The preterm group included 63 children born before a gestational age of 33 weeks, 29 of these children had retinopathy of prematurity (ROP) during the neonatal period, and nine of the children had neurological complications. The control group included 54 children born at term.

Results:
- In the preterm group the RA was significantly smaller compared to the control group. The DA and CA did not differ between the groups. DA and RA correlated with gestational age in the preterm group but no relation with visual acuity, birthweight, ROP or neurological complications was found.

Conclusion: Preterm birth affects the rim of the optic nerve in school-aged children. Since complications of prematurity, like ROP and neurological complications, did not correlate to smaller RA in the preterm group, it is possible that the preterm birth per se affects the visual pathways causing this change in the optic nerve.
Acute sickle cell orbitopathy masquerading as orbital cellulitis

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Introduction: Sickle cell orbitopathy is a rare manifestation of sickle cell crisis and can closely mimic orbital cellulitis, both clinically and radiologically.

Methods and Results: We present a case of a seven-year-old boy with known sickle cell disease (HbSB0) who presented with acute right periorbital swelling, limitation of upgaze, fevers and arm pain. Provisional diagnosis of subperiosteal abscess was made based on computed tomography (CT) scan of the orbits and he was commenced on intravenous antibiotics. Due to minimal clinical improvement, imaging was re-evaluated and revised diagnosis of sickle cell orbitopathy was made. Treatment with hyperhydration resulted in complete resolution of the periorbital swelling and limitation of upgaze within days.

Conclusion: Involvement of the orbital bones in an acute vaso-occlusive crisis is an uncommon manifestation of sickle cell disease and differentiating orbital bone infarction from osteomyelitis and orbital cellulitis clinically as well as radiologically is often very difficult. Although CT is readily accessible in an acute setting, the changes shown may be subtle and misinterpreted. Magnetic resonance imaging (MRI) should be considered as it can demonstrate bone marrow infarction associated with sickle cell orbitopathy more clearly.

Most cases of acute sickle cell orbitopathy resolve with no adverse effects if correctly treated, highlighting the importance of considering it in the differential diagnosis of acute periorbital swelling in children of African descent or those with known history of sickle cell disease.

Non-surgical management of craniofacial dystopia through specialist scleral contact lenses in combination with prismatic glasses

Patsy Terry, Varajini Joganathan, Jennifer McMahon, Steve Wall, Jonathan H. Norris
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Introduction: Surgical rehabilitation of craniofacial dystopia can be challenging. We demonstrate the effective use of specialist scleral contact lenses in combination with prismatic spectacle to correct misalignment of the globe in a paediatric case.

Methods and Results: A 14 year old female with Goldenhaar’s syndrome and right ambylopic esotropia has previously undergone multiple procedures for upper eyelid coloboma and craniofacial clefting. An oblique Fresnel prism successfully corrected the residual vertical misalignment, and a cosmetic scleral contact lens has improved the appearance of the congenital esotropia. The steps involved in the fitting of the scleral lens in this case are demonstrated.

Conclusion: Prismatic spectacle lenses have a successful role in craniofacial dystopia. These can be combined with scleral contact lenses to provide a superior treatment for patients not suited for surgery.
Complications of retained intraorbital wooden foreign body

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**Purpose:** Intraorbital wooden bodies have a high incidence of potentially sight-threatening and life-threatening complications. Besides, intraorbital wood is often not detected by standard diagnostic tests like the computed tomography, so sometimes it is difficult to make correct diagnosis. We report a case of a missed intraorbital wooden body, which was removed 1.5 months after trauma.

**Methods:** 15-year-old boy was admitted to our Clinic due to diagnosis of nodule of the upper eyelid of his right eye, which appeared one week ago. 1.5 months earlier he was struck in the face by a branch during a bicycle ride and he was treated with antibiotic ointment for seven days. On examination there was eyelid granuloma, divergent strabismus, diplopia and eyelid edema. Computed tomography of the orbit showed a retained foreign body along medial orbital wall and the medial rectus muscle.

**Results:** Patient was operated and 3.5 cm piece of wood was removed from the orbit. Postoperatively, he was treated with oral antibiotics following which there was residual restrictive strabismus.

**Conclusions:** Intraorbital foreign bodies may often present a confusing clinical picture. It is imperative to seek the history of trauma in all such cases, regardless of the interval between the trauma and current presentation.

Early and late histological and ultrastructural findings in resected infantile capillary haemangiomas following treatment with topical beta-blocker timolol maleate, 0.5%

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Histopathology Department, Royal Hallamshire Hospital, Sheffield - UK

**Introduction:** Infantile capillary haemangiomas (IH) affect approximately 4-5% of infants. The systemic non-selective β-adrenergic antagonist, propranolol, has become the standard first line treatment for severe IHs. The topical β-antagonist, timolol maleate, has also demonstrated efficacy and safety in treating superficial and some deep capillary haemangiomas. Despite their therapeutic success and prevalent use, the mechanism of action of β-adrenergic antagonists in the treatment of IH is not well understood.

**Methods:** Histopathological, immunohistochemical and electron microscopic evaluation of two periocular IHs excised at one week and 24 months following topical timolol treatment was performed.

**Results:** Distinct morphological differences were observed between spontaneously regressed and β-antagonist treated IHs. The former was characterised by diffuse collagen deposition and interstitial fibrosis, while the latter showed organised concentric collagen IV deposition within obliterated vessel lumen, suggestive of waves of endothelial cell apoptosis, resulting in layers of basement membrane deposits as a stress response. We found evidence of apoptosis on light and electron microscopy as early as one week after the initiation of topical timolol treatment, indicating that apoptosis occurs early in treatment in addition to late during regression.

**Conclusion:** We present for the first time the light and electron microscopic features of IH treated by topical β-antagonist therapy with timolol maleate, 0.5%. We found evidence of endothelial cell apoptosis in IH at one week after initiation of therapy in support of an early apoptotic mechanism for endovascular occlusion and regression in a timolol-treated specimen.
List of Exhibitors

Santhera  L1  
Heidelberg  L2  
Spectrum  L3  
Leica  L4  
MetroVision  L5  
Core Surgical  L6  
Bausch + Lomb  L7  
Albinism and Aniridia Europe  L8
By car - private vehicles between 7.30am & 6.30pm. To avoid the city-centre, approach via the A4142 southern ring-road and take the A4144 Abingdon Rd northwards for about 1.5 miles to St Aldate’s.

By train - Station, via Park End St, New Rd, Queen St and St Aldate’s.

Christ Church, off Oxpens Rd. The nearest park-and-ride - Multi-storey parking is available to the west of Parking car park is about 1.5 miles to the south of Christ Church, off Abingdon Rd.

off George St. Christ Church is a 10 minute walk from the coach station, via George St, Cornmarket St and St Aldate’s.

By coach coach station, via George St, Cornmarket St and St Aldate’s.

Christ Church is a 15 minute walk from Oxford.
Leber’s hereditary optic neuropathy (LHON) is a maternally-inherited, mitochondrial, progressive and rare condition. LHON causes rapid painless central vision loss, and leads to blindness in most cases if left untreated.1,2

**RED FLAGS for suspicion of LHON**3-5

- Usually male
- 15-35 years
- Rapid painless vision loss
- Central or centrocaecal scotoma
- One eye affected initially, followed by the second eye within weeks to months
- Optic disc pseudo-oedema and retinal nerve fibre layer thickening
- Family history of vision loss or LHON
- Non-responder to glucocorticoids

Visit LHONaware.com or email info@LHONaware.com for more information

Abbreviation:
LHON = Leber’s hereditary optic neuropathy

References:
Our goal is to reawaken healthy biologic processes and spark a transformation for people affected by genetic diseases. We are working to address a range of debilitating genetic diseases, including inherited retinal diseases (IRDs), liver-mediated diseases, such as hemophilia, and neurodegenerative diseases. Our approach to gene therapy is to investigate potential treatments that go to an inherited disease at its root by augmenting, replacing or suppressing the function of a mutated gene.
44th Annual Meeting of the European Paediatric Ophthalmological Society

Date and Venue: 7-9 September 2018 Budapest, Hungary

Main topic: Imaging in paediatric ophthalmology

Local hosts: Erika Maka, Krisztina Knézy

https://www.epos-focus.org/meetings